# A Prospective Study Assessing Cancer Patients' Perceptions Regarding the Value of Cancer Genetic Counseling

by

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Hereditary Breast and Ovarian Cancer syndrome (HBOC) is a genetic condition that increases the risk for breast, ovarian, and other cancers. Due to the hereditary nature of this condition, genetic services are offered to patients with these types of cancer to help identify if a hereditary component exists. Several studies have previously investigated the attitudes of patients diagnosed with cancer towards genetic counseling and testing; however, recently diagnosed cancer patients have not received similar attention and their perspective towards the value of genetic testing has not been well studied. To fill this gap, this study was designed to further explore the perspective of patients recently diagnosed with breast or ovarian cancer, mainly at the UPMC Magee Women's Hospital, regarding the value of genetic counseling and to assess their satisfaction levels with the services they received. An initial and a follow-up survey were administered to assess patients' perspectives and experiences with these services after the initial visit and four weeks later to determine if patients saw these services as valuable and satisfactory. Patients were highly satisfied with the genetic counseling services and indicated that it was beneficial and helpful. Patients also saw the genetic counselor as an advocate, support system, and information provider that helped them make medical decisions, including genetic testing. The results of the study are important in that it promotes programs that offer genetic counseling services to conduct internal assessments to gain insight into their patients' levels of satisfaction with the services they are receiving. By actively seeking patients' feedback and addressing concerns,

programs can adapt and enhance their services to better cater to the needs and interests of their patients, thus improving public health.

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## Preface

This program has been one of the most challenging and exciting adventures I have embarked on so far and I would not have been able to reach this point without the support of many. I want to give a special thanks to the program leadership, Jodie Vento, Dr. Andrea Durst, and Dr. Robin Grubs for their unwavering support and dedication to making my dream of becoming a genetic counselor possible. I also wanted to thank my thesis committee, Dr. Andrea Durst, Dr. Phuong Mai, Dr. Jenna Carlson, and Melissa Bourdius for lending me their expertise, mentoring, and assisting me with my thesis project. Also, I want to give a heartfelt gratitude to all my professors, supervisors, and the department of Human Genetic who dedicated their time and effort to educating me and encouraging me ever step of the way.

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# 1.0 Introduction

Cancer genetic counselors have been a resource for patients who have been diagnosed with cancer and have been determined to be at increased risk for hereditary cancer predisposition. Genetic counselors provide patients with valuable information that can be beneficial to patients throughout their treatment process. Additionally, they educate patients about available genetic testing opportunities and help them make sense of test results. Furthermore, cancer genetic counselors assess patients' risk for developing cancer and work together with the patient to form a plan that suits their goals and needs. Moreover, cancer genetic counselors identify other members in the family who can be at an increased risk for cancer and work together with the patient to identify all family members who can benefit from a genetic evaluation. Aside from being an information provider, cancer genetic counselors play a major role in being a support system and advocates for their patients as well as addressing any psychosocial issues associated with a cancer diagnosis, test results, and increased risk for cancer. In the setting of urgent cancer genetic counseling, genetic counselors play a vital role in coordinating genetic testing, discussing risk management and prevention strategies, and supporting and empowering patients in difficult and challenging times. They also collaborate with oncologists, surgeons, and other healthcare providers to ensure comprehensive care for patients.

Several studies have documented the impact cancer genetic counseling had on patients and reported patients' satisfaction and experiences with these services (Bjorvatn et al., 2007; DeMarco et al., 2004; Jagsi et al., 2015; Kausmeyer et al., 2006; Oberguggenberger et al., 2016; Shiloh et al., 1990; Tercyak et al., 2004; Wevers et al., 2012, 2017; E. M. Zilliacus et al., 2010). For example, Kausmeyer et al. conducted a study in the Penn State Cancer Genetics Program that measured

patients' satisfaction with genetic counselors. The results of the study demonstrated an overall satisfaction with the services provided as well as the competency and empathy of the genetic counselor (2006). Another study, conducted by DeMarco et al., tested the reliability of the Genetic Counseling Satisfaction Scale (GCSS), which is a survey designed to measure patient satisfaction with genetic counseling. This study included women who were undergoing counseling and testing for breast and ovarian cancer. The results confirmed the validity of the GCSS and indicated participants' satisfaction with cancer genetic counseling (2004). An additional study conducted by Oberguggenberger et al. looked at psychosocial outcomes and patient satisfaction with genetic counseling and testing. The study demonstrated an overall (84%) patient satisfaction with genetic counseling and testing, and their results were congruent with the study conducted by DeMarco et al. Overall, studies have demonstrated that patients diagnosed with cancer who are receiving genetic counseling have been satisfied with the services provided.

This study aims to measure the perception of urgent referral patients who were recently diagnosed with cancer regarding the value of cancer genetic counseling. The patient population that is included in the study is patients recently diagnosed with cancer who are referred for an urgent genetic counseling appointment for genetic counseling/testing for the purpose of treatment decision making.

Although several studies discussed the perceptions patients with cancer have regarding cancer genetic counseling, information regarding the perceptions of urgently referred patients recently diagnosed with cancer towards genetics services is limited. Genetic counselors play a vital role in counseling urgently referred patients recently diagnosed with cancer as they coordinate genetic testing, discuss risk management and prevention strategies, and support and empower patients in a difficult and challenging times. They also collaborate with oncologists, surgeons, and

other healthcare providers to ensure comprehensive care for patients. Therefore, understanding the perspective of patients receiving these services is needed. This study utilized a survey that was distributed to patients after their initial appointment with the genetic counselor and followed up with a similar survey 4 to 6 weeks from the initial visit to determine if responses have changed. The survey employed questions from The Genetic Counseling Satisfaction Scale (GCSS) and researcher developed questions.

## 1.1 Specific Aims

The following are the two aims of this project:

1. Create a Qualtrics survey utilizing The Genetic Counseling Satisfaction Scale (GCSS) and researcher developed questions to measure opinions of patients newly diagnosed with cancer undergoing genetic counseling and testing on the value of the services they receive.
2. Assess the perception of patients newly diagnosed with breast/ovarian cancer regarding the value of genetic counseling and testing.
    a. Exploring patients' attitudes immediately after the initial session and after a 4-to-6-week period by providing them with an initial and a follow up survey

## 2.0 Manuscript

### 2.1 Background

Hereditary Breast and Ovarian Cancer syndrome (HBOC) is a genetic condition that primarily increases the risk for breast and ovarian cancer, although it can elevate the risk of pancreatic, prostate, and other cancers (Yoshida, 2021). HBOC is an autosomal dominant condition most often caused by a germline pathogenic variant in *BRCA1 or BRCA2*. Several other genes have been established to be associated with an increased risk for breast and/or ovarian cancer including *ATM, PALB2, TP53, STK11, PTEN, CDH1, CHEK2, RAD51C, and RAD51D*. Normally, these genes function as tumor suppressors and are essential components of the double-strand break (DSB) repair mechanism. Pathogenic variant in these genes disable their function, leading to a failure in repairing damaged DNA, therefore, resulting in potential tumor development (Gorodetska et al., 2019). Individuals with a *BRCA1 or BRCA2* mutation have a significantly higher risk for developing certain cancers. For instance, women with mutations in these genes have a 60 to 80% risk of developing breast cancer and 11-53% risk of developing ovarian cancer. They also have a 50 to 65% risk of developing secondary primary breast cancer within 20-45 years of the first breast cancer diagnosis (Kobayashi et al., 2013). Figure 1 visualizes cancer risk for *BRCA1/2* pathogenic variant carriers compared to the risk in the general population (Ferla et al., 2007).

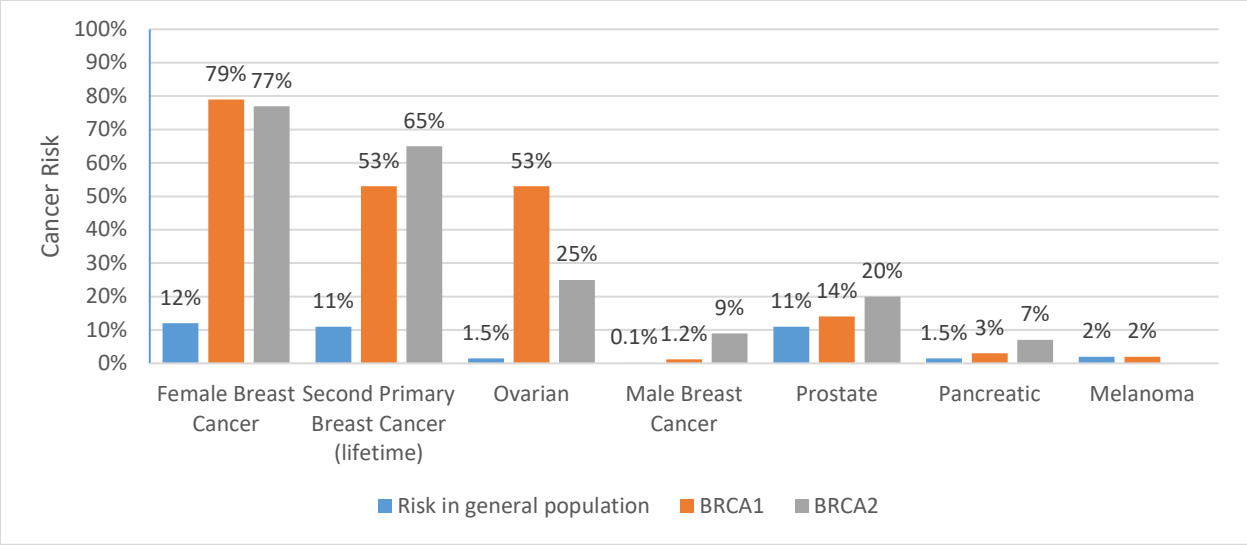


**Figure 1**

**This figure illustrates cancer risk for individuals with BRCA1/2 mutation compared to the general population**

Genetic testing plays a crucial role in identifying patients with hereditary forms of cancer, such as HBOC. Furthermore, it contributes valuable information that can influence the personalization of a patient's treatment plan. Several studies have investigated patients' perspectives and beliefs regarding the benefits and risks of genetic testing (Bjorvatn et al., 2007; DeMarco et al., 2004; Jagsi et al., 2015; Kausmeyer et al., 2006; Oberguggenberger et al., 2016; Shiloh et al., 1990; Tercyak et al., 2004; Wevers et al., 2012, 2017; E. M. Zilliacus et al., 2010). For instance, a qualitative study was conducted among patients diagnosed with breast cancer who were offered treatment-focused genetic testing (TFGT) and those who were not offered TFGT. The results of the study indicated that both groups welcomed genetic testing, with the majority desiring testing soon after diagnosis. However, some expressed concerns about delaying treatment to wait for test results. Participants in both groups highlighted the advantages of genetic testing, which included its impact on medical management and treatment options, as well as providing

information about family risk and prevention strategies for unaffected family members. Overall, participants indicated that the benefits of genetic testing outweigh the risks (E. Zilliacus et al., 2012). When examining factors that positively influence genetic testing for breast and ovarian cancer, studies have shown that gaining knowledge about risk information for breast and ovarian cancer, providing risk information to family members, tailoring the treatment plan, gaining peace of mind, and seeking guidance on prophylactic mastectomy and oophorectomy decisions were the top motivators for choosing *BRCA1/2* testing (Lerman et al., 1994; E. Zilliacus et al., 2012). Conversely, concerns about job discrimination, life insurance discrimination, and adding to the already elevated levels of anxiety were the primary reasons for forgoing cancer genetic testing (Armstrong et al., 2000; E. Zilliacus et al., 2012).

Multiple studies have examined patient satisfaction with genetic counseling and testing (Bjorvatn et al., 2007; DeMarco et al., 2004; Jagsi et al., 2015; Kausmeyer et al., 2006; Oberguggenberger et al., 2016; Shiloh et al., 1990; Tercyak et al., 2004; Wevers et al., 2012, 2017; E. M. Zilliacus et al., 2010). For instance, Weavers et al. aimed to evaluate the outcomes of Rapid Genetic Counseling or Testing (RGCT) in patients newly diagnosed with breast cancer (2012). One of their primary objectives was to gauge patients' satisfaction with RGCT. The study results showed a satisfaction rate of more than 90% for RGCT with 75% of patients recommending RGCT to other patients in their situation (Wevers et al., 2012). DeMarco et al. conducted a study that aimed to assess the reliability and soundness of the Genetic Counseling Satisfaction Scale (GCSS) in the cancer setting and determine the satisfaction levels for women undergoing pre-test cancer genetic counseling for hereditary breast and ovarian cancer. The GCSS is a validated instrument designed to measure patient satisfaction with genetic counseling that was initially designed for the prenatal setting (2004). Researchers successfully demonstrated the reliability of the GCSS and

highlighted the high satisfaction rate among participants regarding their genetic counseling experience (DeMarco et al., 2004). Oberguggenberger et al. also looked at patient satisfaction with cancer genetic counseling, and the results revealed that 84% of patients reported being satisfied or highly satisfied with the services received (2016). Tercyak et al. studied the influence of several personal traits, such as distress, cancer specific distress, personality, and family dynamics on the levels of satisfaction with breast and ovarian cancer genetic counseling (2004). The results showed that there was a significant positive correlation between optimism and satisfaction with genetic counseling and between family function and satisfaction with genetic counseling. The authors did not find a significant difference in the levels of satisfaction when comparing the satisfaction levels in the pre-test and the disclosure sessions. However, the two main factors that were significantly and negatively associated with satisfaction levels were distress in general and distress associated with cancer. Lastly, the results of the study indicated that higher family function increased the likelihood of pre-test satisfaction by 6% (Tercyak et al., 2004).

The previously discussed studies provide insight into patient's attitudes, beliefs, and satisfaction levels regarding genetic counseling and testing. Overall, patients who were previously diagnosed with cancer exhibited positive attitudes and high satisfaction rates with genetic counseling services; however, information about the experiences and perceptions of urgently referred patients recently diagnosed with cancer regarding these services is limited. Therefore, this study aims to fill this knowledge gap and contribute insight into the perspective and attitudes of patients newly diagnosed with breast/ovarian cancer regarding genetic counseling and testing. The study utilized the Genetic Counseling Satisfaction Scale (GCSS) and research team-developed questionnaires to assess patients' perspective regarding the usefulness of genetic counseling in their treatment journey. Additionally, the study aimed to ascertain patients' level of comfort and

satisfaction with genetic counseling services provided to them. Lastly, the study sought to explore patients' attitudes toward genetic counseling services after 4-6 weeks have passed since their initial visit to confirm their initial opinions.

## 2.2 Methods

### 2.2.1 Study Participants

This study recruited patients who were 18 years and older, recently diagnosed with cancer, urgent referral, and seeking in-person cancer genetic counseling at Magee Womens Hospital in Pittsburgh, Pennsylvania. Recruitment began from November 13, 2023, until February 17, 2024, and took place at Magee Womens Hospital after the genetic counseling visit. Patients who agreed to participate in the study had the option of scanning a QR code or using an iPad that was provided to them by the genetic counselor to complete the survey after the appointment while still in clinic. Participants were given the option of how to receive their follow-up survey in 4 to 6 weeks, which included mail, text message, or email. Participants who did not answer any of the survey questions were excluded from the study.

### 2.2.2 Study Design and Data Collection

The Study was approved by the Institutional Review Board of the University of Pittsburgh on November 2nd, 2023. The approval letter is available in Appendix A. This prospective study utilized two surveys for data collection. Additional data was abstracted from participants' medical

records. Both surveys were designed in Qualtrics by utilizing the Genetic Counseling Satisfaction Scale (GCSS) (DeMarco et al., 2004) and researcher team-developed questions. These surveys included questions that reflect participants' perspective regarding the character of the genetic counselor, value of genetic counseling, and satisfaction with genetic counseling. These surveys included positive-response questions and negative-response questions to help responders give accurate answers, The initial survey was administered to participants after the initial genetic counseling visit. The initial survey included information about the study, a consent form that participants had to sign before taking the survey, and 18 questions. The follow-up survey included information about the study, all 18 questions from the initial survey, and two additional questions. A link to the follow-up survey was sent to participants 4-6 weeks after their initial visit.

The survey questions explored several categories that pertain to patient satisfaction with genetic counseling, and most questions were Likert scale based. Some of the explored categories included patients' perception regarding the benefit of genetic counseling, patient's comfort with the genetic counselor, patient's perception regarding the supportive and educational role of the genetic counselor, and patient's motivators for seeking genetic counseling. All the data from both surveys were stored in and analyzed by Qualtrics. Both surveys are listed in Appendix B of the paper. With patients' consent, additional data including patient's sex, race, cancer diagnosis and stage, genetic test results, and surgical procedures, were abstracted from the participants' medical record, and securely stored on the Pitt One Drive.

### 2.2.3 Data Analysis

Qualtrics was utilized for data analysis regarding the data generated from the surveys, which consisted of descriptive statistics (Qualtrics, Provo, UT). One survey was removed from the

analysis as none of the questions were answered. Additionally, Excel was used to generate tables and graphs from raw data. Data from the medical records were organized manually and were used to create descriptive statistics, including, sex, race, age, cancer diagnosis, and genetic test results. Due to the small sample size, no statistical tests were done.

## 2.3 Results

One hundred and six patients were eligible to participate in the study, however, 70 patients were asked to participate due to patients' time constraints. The initial survey was offered to 70 potential participants, and 41 completed the survey for a response rate of 58.6%. Thirty-eight out of the 41 participants (92.7%) completed the initial survey on the day of the appointment, while 3 participants (7.3%) completed the survey at a different date. The follow-up survey was offered to the 41 participants who completed the initial survey, and 17 completed the follow-up survey for a response rate of 41.5%.

### 2.3.1 Participants Demographics

The demographics of the study participants are summarized in Table 1. The ages of the participants ranged from 37 to 86 with a mean age of 57. All participants were females, 38 identified as "White" (92.7%), 2 identified as "Black" or "African American" (4.9%), and 1 identified as "Asian" (2.4%). No males participated in this study.

**Table 1 Demographics**

| | | Mean | SD | Median | (N=41) N (%) |
|---|---|---|---|---|---|
| Age | | 57.1 | 13.3 | 54 | |
| Sex | Male | | | | 0 (0) |
| | Female | | | | 41 (100) |
| Race | White | | | | 38 (92.7) |
| | Black or African American | | | | 2 (4.9) |
| | Asian | | | | 1 (2.4) |

### 2.3.2 Cancer Diagnosis and Genetic Test Results

Thirty-eight out of the 41 participants were diagnosed with breast cancer (92.7%) and 3 were diagnosed with ovarian cancer (7.3%). Five types of breast cancer were identified in the study sample, with invasive ductal carcinoma (IDC) being the most common (63.2%), followed by triple negative breast cancer and ductal carcinoma in situ (13.2% each), invasive lobular carcinoma (7.9%), and metaplastic carcinoma (2.6%). Cancer diagnoses for the participants are summarized in Table 2. All participants underwent genetic testing. Twenty-eight participants received a negative result (68.3%), 8 received a VUS (19.5%), and 5 received a pathogenic variant (12.2%). Pathogenic variants were identified in 3 genes, *BRCA1* (3 participants), *CHEK2* (1 participant), and *BARD1* (1 participant). Testing results are summarized in Table 3.

**Table 2 Cancer Diagnosis and Genetic Mutations**

| | **Type** | **Initial Survey N=41 (%)** | **Follow-up survey N=17 (%)** |
|---|---|---|---|
| Breast Cancer | | **38 (92.7)** | **16 (94.1)** |
| | IDC | 24 (63.2) | 7 (43.8) |
| | Triple | 5 (13.2) | 2 (12.5) |

| | | | |
|---|---|---|---|
| | Negative | | |
| | DCIS | 5 (13.2) | 4 (25) |
| | ILC | 3 (7.9) | 2 (12.5) |
| | Metaplastic | 1 (2.6) | 1 (6.3) |
| | | | |
| Ovarian Cancer | | **3 (7.3%)** | **1 (5.9)** |
| | | | |
| Genes with pathogenic variant | | | **2 (11.8)** |
| | BRCA1 | | 2 (11.8) |
| | CHEK2 | | 0 (0) |
| | BARD1 | | 0 (0) |

**Table 3 Genetic Testing Results**

| **Testing outcome:** | Initial Survey N (%) | Follow-up Survey N (%) |
|---|---|---|
| Pathogenic variant | 5 (12.2) | 2 (11.8) |
| Variant of Uncertain Significance | 8 (19.5) | 3 (17.6) |
| Negative | 28 (68.3) | 12 (70.6) |

### 2.3.3 Survey Responses

Participants both in the initial and follow-up surveys responded positively to questions that assessed patients' perception of the genetic counselor. This was demonstrated by the mean value for each question, which ranged between 4 and 5, indicating that participants' responses ranged between Strongly Agree and Agree. Respondents had the lowest mean response for question 4 (4.0) in the initial survey and question 5 in the follow up survey. For questions that focused on patients' perceptions regarding the value of genetic counseling and questions that focused on patients' satisfaction with generic counseling services, the means in the initial and follow-up

surveys were between 4 and 5 for positive-response questions and between 1 and 2 for negative-response questions. The mean values indicate that participants' responses ranged between Strongly Agree and Agree for the positive-response questions and between Strongly Disagree and Disagree for the negative-response questions. Table 4 lists the initial and follow-up survey questions, and provides the mean value for the responses, from which interpretations can be drawn.

**Table 4 Survey Responses**

| **Questions focused on patients' perceptions of the genetic counselor** | Initial Survey Means N=41 | Follow-up Survey Means N=17 |
|---|---|---|
| 1. My genetic counselor seemed to understand the stresses I was facing | 4.5[1] | 4.8 |
| 2. My genetic counselor was truly concerned about my well-being | 4.5 | 4.9 |
| 3. My genetic counselor was supportive and respectful of my decisions | 4.7 | 4.8 |
| 4. I felt better about my health after meeting with my genetic counselor | 4.0 | 4.4 |
| 5. My genetic counselor helped me make decisions about genetic testing | N/A[2] | 4.3 |
| 6. My genetic counselor helped me identify what I needed to know to make decisions about what could happen to me | 4.5 | 4.5 |
| **Questions focused on patients' perceptions regarding the value of genetic counseling** | | |
| 7. I believe the counseling that I received has helped me cope better with my cancer diagnosis | 4.4 | 4.2 |
| 8. Having a genetic counselor on my medical team was not helpful[3] | 1.5 | 1.3 |
| 9. The information that I received in the session was not helpful | 1.3 | 1.1 |
| 10. The genetic counseling session was valuable to me | 4.6 | 4.8 |
| 11.The counseling that I received helped me feel more confident making my next medical decisions regarding my cancer diagnosis | 4.4 | 4.5 |

[1] Likert-Scale questions with 1= Strongly Disagree, 2=Disagree, 3=Neutral, 4=Agree, 5= Strongly Agree

[2] N/A are questions that were not included in the initial survey

[3] Questions in red are negative-response questions

| | | |
|---|---|---|
| 12. The information I received made me feel empowered about my health decisions | N/A | 4.3 |
| **Questions focused on patients' satisfaction with genetic counseling** | | |
| 13. The genetic counseling session was about the right length of time I needed | 4.5 | 4.8 |
| 14. I would feel comfortable contacting the genetic counselor to ask them additional questions | 4.7 | 4.7 |
| 15 If I was given the choice again, I would not seek genetic counseling | 1.2 | 1.3 |
| 16. I would not recommend cancer genetic counseling for someone in a similar situation as mine | 1.4 | 1.2 |

For the initial survey, the most common reason for seeking genetic counseling was "Doctor's recommendations", with 92.7% of participants (n=38) selecting that as one of their answers. On the follow-up survey, "Make treatment decisions about my cancer" was the most common reason for seeking genetic counseling, with 88.2% of participants (n=15) selecting that answer. The least common reason for seeking genetic counseling for participants in the initial and follow-up survey was "seeking support", with 9.76% and 11.76% of participants selecting that choice, respectively. Figure 2 represent participants' motivators for seeking genetic counseling for both the initial and follow-up survey, respectively.

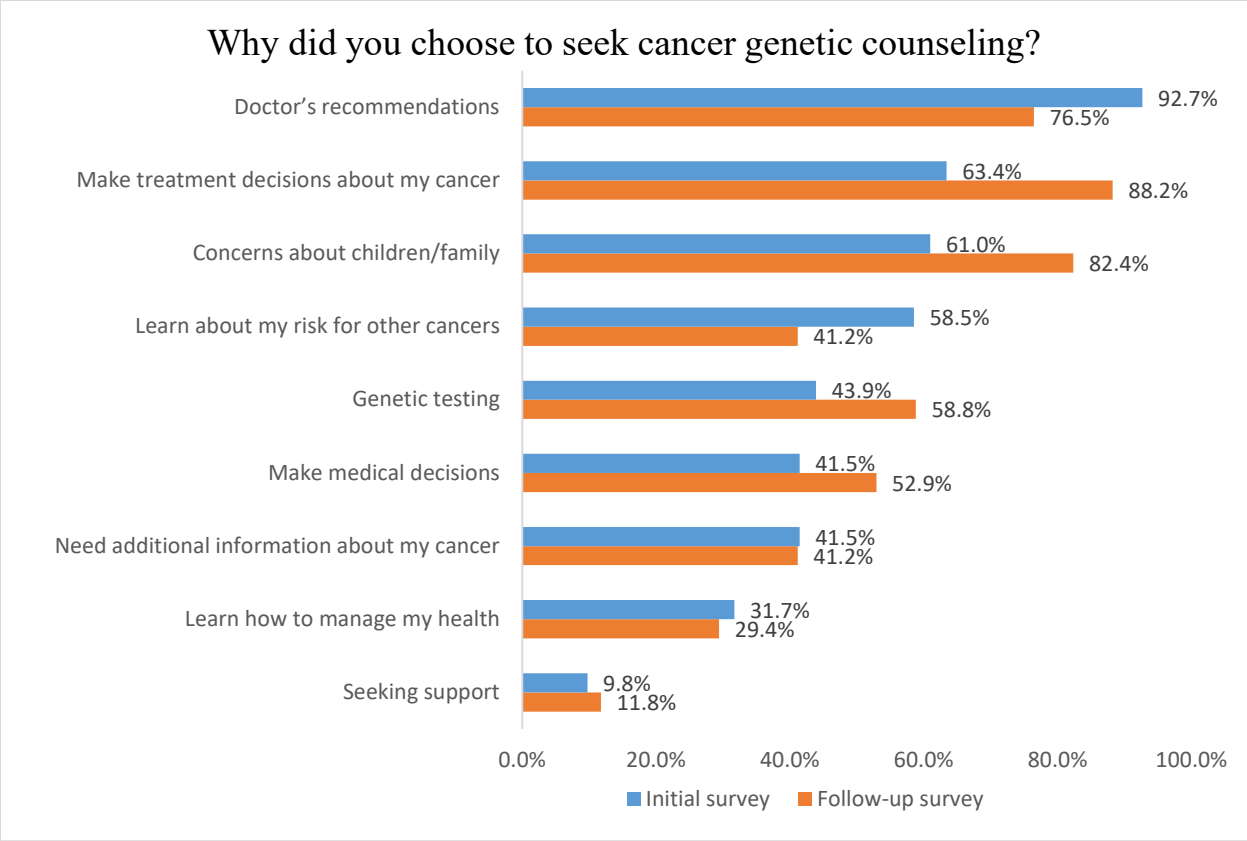


**Figure 2 Motivators for Genetic Counseling.**

**Intial Survey N=41, Follow-up Survey N=17**

When asked about cancer treatment progress, 78.1% (n=32) of participants from the initial survey indicated that they have not started treatment yet, while the remaining 21.9% (n=9) reported being in the middle of completing treatment for their cancer. Looking at participants from the follow-up survey, 64.7% (n=11) of them reported being in the middle of receiving treatment for their cancer, while the remaining 35.3% (n=6) indicated that they have not started treatment yet. No one in either survey indicated having completed cancer treatment.

## 2.4 Discussion

This study was conducted to explore the perspective of patients newly diagnosed with breast or ovarian cancer regarding the value of genetic counseling and to examine their satisfaction levels with genetic counseling services. The results of the study indicated that participants were highly satisfied with the services being provided, viewed it as beneficial and important, and saw the genetic counselor as an empathetic and supportive provider that facilitates healthcare decision making.

Most participants viewed the genetic counselor as an understanding, caring, and considerate healthcare provider who was able to comprehend the stress the patients were experiencing and was concerned about their well-being. Participants agreed that the genetic counselor provided information, guidance, comfort, and support during this process while being respectful of their healthcare decisions. Additionally, the genetic counselor was seen as a facilitator for the medical decision process, including genetic testing. These findings were evident in participants' responses to the survey questions, particularly questions that were designed to gain insight into participant's perspective regarding the affectivity of the genetic counselor. These results are congruent with other studies that examined the interaction between cancer patients and their genetic counselors, such as the study conducted by Kausmeyer et al. (2006), which showed that the overwhelming majority of patients were highly satisfied with the genetic counselors, their attitude, and their ability to provide information, psychosocial support, understanding, and compassion (2006). Being satisfied with the genetic counselor and recognizing their role as information provider, support system, and a patient's advocate, can contribute to the patient's overall satisfaction with the genetic counseling experience. Therefore, genetic counselors must continue to advocate for their patients, provide psychosocial support, and help educate them.

Participants' positive responses to questions that investigated patients' perceptions regarding the value of genetic counseling suggest that patients viewed genetic counseling as a valuable experience. Most patients indicated that genetic counseling helped them cope better with their cancer diagnosis, provided them with helpful and valuable information, gave them confidence to make medical decisions, and empowered them. Other studies have reported similar benefits genetic counseling had on patients. For instance, Yuen et al. conducted a study to determine if cancer genetic counseling had an impact on patient's empowerment levels (2020). The result of their study demonstrated that patients who underwent cancer genetic counseling displayed elevated levels of empowerment (Yuen et al., 2020). Similarly, Axillbund et al. assessed patients perspective regarding the value of genetic counseling for familial pancreatic cancer and found that 93% of participants indicated that genetic counseling was valuable and helpful (2005). The results of this study demonstrate the benefits genetic counseling can have on patients who are diagnosed with cancer, as it provides them with coping strategies, helpful information, empowerment, and may contribute to their overall satisfaction with genetic counseling. Therefore, genetic counseling services must continue to grow and expand to reach the largest number of patients, especially those living in areas where such services are not available.

Satisfaction with the genetic counseling experience was demonstrated by participants' approval of the length of the counseling session, comfort level with reaching back to the genetic counselor with questions, willingness to recommend cancer genetic counseling for people in their situation and agreeing to have genetic counseling if given the choice again. In addition to these factors, the combination of participants' positive views of their genetic counselor and the beneficial outcomes they received from the counseling session can play a role in the overall satisfaction levels among participants. Numerous studies have documented the high satisfaction

levels among patients undergoing genetic counseling. For instance, Bjorvatn et al. conducted a study to further assess satisfaction levels for cancer genetic counseling patients (2007). The results of the study demonstrated high satisfaction rate with genetic counseling for the majority of patients and discovered that patients were able to estimate their cancer risk levels more accurately after receiving genetic counseling compared to before (Bjorvatn et al., 2007). Additional studies such as the ones conducted by DeMarco et al., 2004, Jagsi et al., 2015, Kausmeyer et al., 2006, Oberguggenberger et al., 2016, Shiloh et al., 1990, Tercyak et al., 2004, Wevers et al., 2012, 2017, and E. M. Zilliacus et al., 2010, have all examined patients' satisfaction with genetic counseling and all found high satisfaction levels. Although these studies do not focus on patients who were recently diagnosed with cancer, their findings align with the results of this study, thus lending additional strength and support to the findings in this small study population. The data collected indicated that patients at UPMC Magee Womens Hospital who are newly diagnosed with a breast or ovarian cancer received positive genetic counseling experiences and viewed genetic counseling as an integral part of their healthcare, providing them with beneficial and helpful information that enabled them to make decisions regarding their health. Cancer genetic counseling programs that provide genetic counseling services to patients may benefit from conducting internal investigation to measure the levels of satisfaction among their patients and potentially make any necessary changes to best serve their patients. It is important to note that the completion rate of the follow-up survey between participants who had a pathogenic variant (40%) and those who had a negative (VUS included) test result (41.7%) were similar. This finding combined with participants positive results to the follow-up survey may suggest that testing results did not impact satisfaction with genetic services and participant's positive views regarding the value of genetic counseling and testing. More research is needed to further explore this finding.

### 2.4.1 Limitations

This study has several limitations, the first of which is the small sample size of 41 participants and the lack of completed follow-up surveys, which prevents any statistical comparison. Second, the study only offered participation to patients who were seen in person and excluded telemedicine patients, therefore, failing to capture the experiences of this sub-set of patients. Third, the study sample did not have a fair representation of different racial groups, as it mainly included White females (92.7%), and was drawn from a single institution located in an urban city. Allegheny county is composed of various racial groups including White (Non-Hispanic) (77.4%), Black or African American (non-Hispanic) (12.5%), Asian (Non-Hispanic) (3.92%), Two+ (non-Hispanic) (3.36%), and White (Hispanic) (1.01%) (*Allegheny County, PA,* 2021). By focusing mainly on White females, the study failed to capture the experiences, needs, and perspectives of individuals from other racial groups and ethnic backgrounds, thus making generalizability difficult. Fourth, volunteer bias may have influenced the sample of the study. For instance, it could have occurred that only patients who were satisfied with genetic counseling services and the genetic counselor agreed to participate in the study while patients who were unsatisfied were reluctant to participate, therefore their experiences were not documented. Lastly, the study was not anonymous, and participants completed the survey while the genetic counselor was present, and it is possible that participants may have altered their responses to reflect positive answers rather than answering honestly.

Future studies need to include a more representative sample that encompasses participants from different racial groups and ethnic backgrounds in order to aid in the generalizability of the study. Additionally, future studies should consider including telemedicine patients as they may have different experiences regarding their satisfaction with genetic counseling and how they view

these services. Moreover, studies should consider looking at patients diagnosed with other types of cancer, such as colon, uterine, kidney, and pancreatic and assess their satisfaction with cancer genetic counseling. Lastly, more qualitative studies should be conducted to uncover more details about patients' experiences with cancer genetic counseling.

## 2.5 Conclusion

The goal of this study was to assess the perspective of patients newly diagnosed with breast or ovarian cancer at UPMC regarding the value of genetic counseling and to further understand their satisfaction levels with the services provided. Although the study sample was limited in number, the majority of participants indicated that genetic counseling was a valuable part of their healthcare during this time of uncertainty. Additionally, participants highlighted the support and valuable information they received and indicated its role in facilitating healthcare decisions making. Moreover, the majority of participants expressed high satisfaction with the counseling services and viewed the genetic counselor as an empathetic, information giving, supportive, and respectful healthcare provider who helped them reach an informed medical decision. Understanding patients' perspectives and satisfaction with genetic counseling services is crucial, as it offers valuable insights into how these services are perceived. By actively seeking feedback and addressing concerns, programs can adapt and enhance their services to better cater to the needs and interests of their patients. Therefore, additional qualitative studies with larger sample size and improved inclusivity, are necessary to tease out the nuanced aspects of patient's satisfaction with genetic counseling.

## 3.0 Research Significance to Genetic Counseling and Public Health

The goal of this study was to assess the perspective of urgent care cancer patients regarding the value of genetic counseling and to investigate patients' satisfaction levels with genetic counseling services. The results of the study are significant to the field of genetic counseling and public health alike, as they demonstrate the value of genetic services in patient care and illustrate the importance of service expansion where services are limited.

The current study sheds some light directly on a principal aspect of genetic counseling, which is the service itself, by exploring patients' feedback regarding the value of the counseling services. This is significant to the field of genetic counseling and public health because evaluation of services via patient feedback is a vital component that helps determine if these services are benefiting patients and providing them with information, support, and knowledge that can be useful to them. Genetic counselors provide patients with the risk assessment and tools to help them make informed decisions about cancer screenings and prevention measures; therefore, investigating patients' perspectives regarding these services can provide insight into the efficacy of risk communication and the extent to which patients perceive these services as empowering or distressing. Moreover, understanding patients' perspectives regarding genetic counseling can help in the identification of barriers to accessing genetic counseling services. This leads to initiatives that aim to address these barriers to improve access, reduce health disparities, and promote health equity, which are main pillars of public health. Since helping patients reach an informed decision is a main aspect of genetic counseling, understanding the impact counseling has on the decision-making process and how patients weigh the risks and benefits associated with genetic testing can help genetic counselors tailor their counseling sessions to best help their patients. This is also true

for patients who are urgently referred to genetic counseling and testing, especially that the information they receive, such as genetic test results and risk-reducing strategies can help them and their provider tailor their treatment and management process.

Public health encompasses three primary functions: assessment, policy development, and assurance. These functions are further delineated into ten essential public health services, offering a structured framework to support the core mission and operations of public health initiatives (*CDC,* 2024). One of the 10 essential public health services is "Improve and innovate public health functions through ongoing evaluation, research, and continuous quality improvement". This study embodies this public health service because it is performing this function. It is essential that we evaluate the services we are offering so that we can ensure they meet the goals of the services. The results of this study indicated that patients valued the services and viewed them as informative and influential parts of the decision-making process. In addition, participants regarded the counselor as a compassionate healthcare provider who effectively educated, supported, and assisted them in the decision-making process while respecting their decisions. These findings suggest that the genetic counseling services at UPMC Magee Women's Hospital are beneficial to patients and play a role in helping them reach a medical decision. And thus, this is likely to be an important service for all patients recently diagnosed with cancer who meet criteria for genetic counseling/testing. Additionally, these results reflect the quality of the genetic counselor and their ability to "communicate effectively to inform and educate people about health, factors that influence it, and how to improve it" (*CDC,* 2024), which is another essential public health service. Therefore, this study should be a catalyst for genetic counseling programs that offer services to cancer patients to consider conducting a similar study to further understand how their patients view and value genetic counseling. Additionally, more qualitative studies should be conducted, as qualitative research is

better equipped to gain insight into patients' overall experience. This is important as patients' feedback can contribute to improving genetic counseling services and tailoring such services to serve all patients' needs, thus contributing to the improvement of genetic counseling and public health.

# Appendix A IRB Approval Letter

University of Pittsburgh | Institutional Review Board
Office of Research Protections

## INITIAL APPROVAL (Expedited)
**Continuing Review Not Required**

| | |
|---|---|
| Date: | November 2, 2023 |
| IRB: | STUDY23080139 |
| PI: | Elian Buchi |
| Title: | A Prospective Study Assessing Cancer Patients' Perceptions Regarding the Value of Cancer Genetic Testing |

The Institutional Review Board reviewed and approved the above referenced study. The study may begin as outlined in the University of Pittsburgh approved application and documents. Continuing Review is not necessary under 45 CFR 46.109(f)(1)(i).

**Approval Documentation**

| | |
|---|---|
| Review type: | Initial Study |
| Approval Date: | 11/2/2023 |
| Expedited Category(ies): | (5) Data, documents, records, or specimens, (7)(a) Behavioral research |
| Approved Documents: | • Initial survey, Category: Data Collection;<br>• Follow-up survey, Category: Data Collection;<br>• Consent form, Category: Consent Form;<br>• Study Protocol , Category: IRB Protocol; |

As the Principal Investigator, you are responsible for the conduct of the research and to ensure accurate documentation, protocol compliance, reporting of possibly study-related adverse events and unanticipated problems involving risk to participants or others. The HRPO Reportable Events policy, Chapter 17, is available at http://www.hrpo.pitt.edu/.

Clinical research being conducted in an UPMC facility cannot begin until fiscal approval is received from the UPMC Office of Sponsored Programs and Research Support (OSPARS).

If you have any questions, please contact the University of Pittsburgh IRB Coordinator, Ali Arak.

If you are conducting a federally funded clinical trial, there may be additional requirements. Please refer to information on clinical trials https://www.ecshsr.pitt.edu/ct/documents.

*Please take a moment to complete our* Satisfaction Survey *as we appreciate your feedback.*

The University of Pittsburgh has a Federal Wide Assurance approved through the Office of Human Research Protections (FWA00006790).

## Appendix B Surveys

This section will contain the main questions listed from the initial and follow-up surveys. The majority of questions were Likert-Scale based and participants were to choose from these possible answers: “Strongly agree, Agree, Uncertain, Disagree, Strongly Disagree”, therefore, Likert-Scale questions will not have the possible answers.

**Initial Survey Questions**

1. My genetic counselor seemed to understand the stresses I was facing
2. I believe the counseling that I received will help me cope better with my cancer diagnosis
3. The genetic counseling session was about the right length of time I needed (logic question, if the answer was “Disagree or Strongly Disagree”, this question was asked
    a. Was the session too long or too short (answers: Too long, Too short)
4. Why did you choose to seek cancer genetic counseling? (choose all that apply)
    a. Possible answers: Genetic testing, Doctor’s recommendations, Need additional information about my cancer, Concerns about children/family, Learn how to manage my health, Seeking support, Learn about my risk for other cancers, Make medical decisions, Make treatment decisions about my cancer, Other (please specify)

5. My genetic counselor was truly concerned about my well-being
6. Having a genetic counselor on my medical team was not helpful
7. I would feel comfortable contacting the genetic counselor to ask them additional questions
8. The information that I received in the session was not helpful (Logic question, if the answer was Agree or Strongly Agree, this question was asked)
    a. You indicated that the information you received during the session was not helpful. Please share your thoughts and comments about that
9. My genetic counselor was supportive and respectful of my decisions
10. The genetic counseling session was valuable to me
11. I felt better about my health after meeting with my genetic counselor
12. I would not recommend cancer genetic counseling for someone in a similar situation as mine (Logic question if the answer was Agree or Strongly Agree, this question was asked)
    a. You indicated that you would not recommend cancer genetic counseling for someone in a similar situation as you. Please share your thoughts on why you would not recommend genetic counseling
13. f I was given the choice again, I would not seek genetic counseling
14. My genetic counselor helped me identify what I needed to know to make decisions about what could happen to me
15. The information I received made me feel empowered about my health decisions
16. Have you started treatment for your cancer (Options included)
    a. No, I have not started treatment for my cancer

b. I'm in the middle of completing treatment for my cancer (Chemotherapy, surgery, radiation)
c. I have completed my treatment for cancer

**Follo-up Survey Questions**

1. My genetic counselor seemed to understand the stresses I was facing
2. I believe the counseling that I received will help me cope better with my cancer diagnosis
3. The genetic counseling session was about the right length of time I needed (logic question, if the answer was "Disagree or Strongly Disagree", this question was asked
    a. Was the session too long or too short (answers: Too long, Too short)
4. Why did you choose to seek cancer genetic counseling? (choose all that apply)
    a. Possible answers: Genetic testing, Doctor's recommendations, Need additional information about my cancer, Concerns about children/family, Learn how to manage my health, Seeking support, Learn about my risk for other cancers, Make medical decisions, Make treatment decisions about my cancer, Other (please specify)
5. My genetic counselor was truly concerned about my well-being
6. Having a genetic counselor on my medical team was not helpful
7. I would feel comfortable contacting the genetic counselor to ask them additional questions
8. The information that I received in the session was not helpful (Logic question, if the answer was Agree or Strongly Agree, this question was asked)

    a. You indicated that the information you received during the session was not helpful. Please share your thoughts and comments about that

9. My genetic counselor was supportive and respectful of my decisions
10. The genetic counseling session was valuable to me
11. I felt better about my health after meeting with my genetic counselor
12. I would not recommend cancer genetic counseling for someone in a similar situation as mine (Logic question if the answer was Agree or Strongly Agree, this question was asked)
    a. You indicated that you would not recommend cancer genetic counseling for someone in a similar situation as you. Please share your thoughts on why you would not recommend genetic counseling
13. f I was given the choice again, I would not seek genetic counseling
14. My genetic counselor helped me identify what I needed to know to make decisions about what could happen to me
15. The information I received made me feel empowered about my health decisions
16. Have you started treatment for your cancer (Options included)
    a. No, I have not started treatment for my cancer
    b. I'm in the middle of completing treatment for my cancer (Chemotherapy, surgery, radiation)
    c. I have completed my treatment for cancer
17. My genetic counselor helped me make decisions about genetic testing
18. The counseling that I received helped me feel more confident making my next medical decisions regarding my cancer diagnosis

19. Did you choose to do genetic testing (Options include:)

    a. Yes

    b. No

    c. Still deciding

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