Spring 2021

Assessing the Anticipated Needs of Transgender Patients In Cancer Genetic Counseling

Jacqueline Baquet

ASSESSING THE ANTICIPATED NEEDS OF TRANSGENDER PATIENTS IN CANCER GENETIC COUNSELING

by

Jacqueline Baquet

Bachelor of Science
University of South Carolina, 2019

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:

Diane Koeller, Director of Thesis

Peyton Nunley, Reader

Kristl Tomlin, Reader

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

ABSTRACT

Most cancers are sporadic, but 5-10% of all cancer is hereditary, or caused by a heritable genetic mutation. A patient's medical history, family history, genetic test results, intact organs (e.g., ovaries) at an increased risk for developing cancer, and the availability and accessibility of interventions are used to make recommendations for cancer-risk management. In addition to basic medical care, transgender patients have healthcare needs that differ from those of cisgender patients such as expert care related to using hormones or having gender-affirming surgery, as well as unique mental health concerns. Transgender individuals may also experience a greater number of barriers to accessing care than cisgender individuals.

The purpose of this study was to explore the motivations and needs of transgender individuals who may seek cancer genetic counseling. We aimed to determine where current practices could be improved to increase comfortability and inclusivity of transgender patients. Eighty-seven transgender individuals participated in an online questionnaire that asked about their personal perspectives on comfort and preferences regarding current genetic counseling practices.

Most participants reported that they would feel comfortable sharing their pronouns, hormone therapies, and surgical history on an intake form before their genetic counseling appointment. The results suggested that comfort levels between the different current practices regarding pedigree nomenclature had no statistical differences, although most participants would not be comfortable being represented as their sex assigned at birth on

a pedigree. When assessing motivations, evidence demonstrated that most participants would want to discuss how hormone and surgical therapies could impact personal cancer risk. These findings reinforce recommendations from existing literature regarding the adaptation and evolution of current practices to meet the needs of transgender patients while highlighting the need for standardized training in order to provide comprehensive, inclusive care for all patients, regardless of gender identity.

TABLE OF CONTENTS

LIST OF TABLES

LIST OF FIGURES

CHAPTER 1

BACKGROUND

1.1 Introduction to Hereditary Cancer

Cancer is one of the most prevalent diseases worldwide. The National Cancer Institute (NCI) is the principal agency for conducting and supporting cancer research, prevention, diagnosis, and treatment in the United States. The NCI has been documenting cancer incidence data in the Surveillance, Epidemiology, and End Results (SEER) Program since 1973 and makes the data publicly accessible online. According to SEER, there were 1,806,590 new cases of cancer in 2020. While most cancers are sporadic, approximately 5-10% of all cancer is hereditary – it occurs in a person with a heritable genetic mutation in a cancer predisposition gene (Dalton et al., 2014). When an individual has a genetic predisposition to cancer, they are at an increased risk for developing certain types of cancer depending on which gene has a mutation. Hereditary cancers tend to occur at younger ages than sporadic cancers and individuals with a hereditary cancer syndrome can have multiple cancers in their lifetime.

Based on these risks, the National Comprehensive Cancer Network (NCCN) has published guidelines for genetic testing for hereditary cancer syndromes and cancer risk management in individuals who have hereditary cancer predisposition syndromes. These recommendations for cancer screenings and preventative therapies aim to reduce the probability that cancer will occur or reoccur in these individuals. For example, the NCCN Guidelines for Genetic/Familial High-Risk Assessment for Breast, Ovarian, and

Pancreatic cancers include criteria for factors in an individual's personal and family medical history make them an appropriate candidate for genetic counseling and genetic testing. Some of these factors include being diagnosed with breast cancer under 46-years-old, metastatic prostate cancer at any age, or having a blood relative with a known mutation in a cancer susceptibility gene (NCCN 2020).

If someone is found to be at an increased risk for developing cancer based on their family history or their genetic testing results, the NCCN recommends earlier and increased cancer surveillance. Some of these recommendations include starting breast magnetic-resonance imaging (MRI), mammograms, colonoscopies, endoscopies, and other screenings at younger ages or undergoing these cancer screenings more frequently in order to identify cancers earlier and optimize any necessary treatment.

1.2 Genetic Counseling and Testing for Cancer Predispositions

Physicians use the NCCN guidelines as well as other recommendations from professional societies such as the American Cancer Society, American College of Obstetricians and Gynecologists, and the American College of Medical Genetics and Genomics to determine which of their patients need to be referred to a genetic counselor, based on that individual's personal and family history.

Genetic counselors are healthcare professionals with training in both medical genetics and psychosocial counseling. A cancer genetic counselor specializes in evaluating patients for hereditary cancer syndromes. In a typical cancer genetic counseling session, the genetic counselor elicits a patient's personal and family medical history and conducts a risk assessment to determine the likelihood that a patient's cancer or the cancer in their family may be caused by a genetic predisposition syndrome. The genetic counselor then

communicates that risk assessment to the patient, facilitates decision making regarding options for genetic testing, educates the patient on cancer surveillance and prevention, and arranges genetic testing when appropriate (Schneider, 2011). During a genetic counseling session, the counselor also explores psychosocial issues regarding motivations for testing, feelings about the risk information, and overall emotional state of the patient.

When eliciting a patient's family history, the genetic counselor will document the information as a pedigree, or a diagram of the family tree. Once the pedigree is drawn, the genetic counselor looks for patterns that are consistent with those seen in hereditary cancer syndromes and presents this information to the patient as a probability that they could have a hereditary cancer syndrome. The goal of providing this information is to aid in the patient's decision making about genetic testing and future medical management. If a patient decides to pursue genetic testing, the genetic counselor helps them with decisions regarding appropriate testing and follow-up (Glessner et al., 2012).

The National Society of Genetic Counselors (NSGC) has established Practice Guidelines for Human Pedigree Nomenclature. The purpose of these guidelines is to create a standardized way to accurately and appropriately represent individuals, their relationships, and their medical status for risk assessment and future reference for other medical providers (Bennett et al., 2008). Traditionally, based on the binarization of gender in most Western cultures, females and males have been represented by circles and squares, respectively. If an individual's gender is unknown, they may be represented by a diamond (Bennett et al., 2008; Sutherland et al., 2020).

The most recent update to these guidelines was in 2008, which recommended a standardized way of representing transgender individuals. However, periodic revision of

these guidelines is imperative to ensure inclusivity of all patients (Sheehan et al., 2020). The NCCN and NSGC guidelines on how to represent transgender individuals on a pedigree differ and there is currently no universally accepted and standardized way to represent gender identity and sex assigned at birth. The lack of consensus creates confusion between medical professionals and does not validate the gender identity of transgender patients. There have been suggestions to create a unique symbol for transgender and nonbinary patients within the field of genetic counseling, which underlines the need for standardization and reform of the current practice guidelines (Sheehan et al., 2020; Tuite et al., 2020).

1.3 Overview of Transgender Healthcare

A person's sexuality and gender are made up of many features including gender identity, gender expression, sex assigned at birth, and sexual orientation. Several transgender education and advocacy organizations have published infographics to explain these aspects of the gender and sexuality spectrums. To describe gender identity, three independent spectrums are used to measure the degree to which someone identifies with the female gender, male gender, or other genders (Coleman et al., 2012). Gender expression is the degree to which someone outwardly expresses themselves in a feminine, androgenous, or masculine way. In the United States, children are typically assigned male, female, or intersex at birth before their gender identity is developed, which is known as "sex assigned at birth" (Puechl et al., 2019).

According to the Centers for Disease Control and Prevention's (CDC) 2018 Behavioral Risk Factor Surveillance System (BRFSS), over 1.4 million Americans identify as transgender and/or nonbinary. The term "transgender" is used to describe

people whose gender identity does not align with their sex assigned at birth. “Cisgender” is a term used to describe individuals whose gender identity does alighn with their sex assigned at birth. Someone who is transgender may identify with the female gender, male gender, another gender, no gender, or a combination of genders. Someone who does not identify with the female or male genders may describe their gender as “nonbinary” (Rafferty, 2018). When a person’s gender does not align with their sex assigned at birth, they may choose to express their gender through social changes, hormonal therapies, and/or surgical transitions.

The World Professional Association for Transgender Health (WPATH) published Standards of Care for the Health of Transsexual, Transgender, and Gender-Nonconforming People. These guidelines describe therapies available to aid in the transition from sex assigned at birth to true gender, including hormonal and surgical options. Hormone therapy is the use of hormones to induce more feminine or masculine secondary sex characteristics. Changes that occur in individuals who are prescribed gender-affirming hormones typically occur over the course of two to five years. There are some possible risks to taking these hormones, including a potential increase in cancer risk, although much of the data is minimal and sometimes conflicting (Sutherland et al., 2020).

Gender-affirming surgeries may involve altering an individual’s breast tissue, ovaries, and genitalia. Breast and chest reconstruction may be referred to as “top surgery” and typically involves either a breast augmentation mammoplasty or a subcutaneous mastectomy with chest contouring (Coleman et al., 2012). Subcutaneous, or nipple-sparing mastectomies, are not typically prophylactic bilateral total mastectomies, which is

the recommended surgical prevention for some high-risk hereditary breast cancer syndromes (Guillem et al., 2020). Additional surgical transitions may include "bottom surgery," or surgery involving reproductive and/or genital organ reconstruction or removal (Coleman et al., 2012). Surgery to remove reproductive organs such as the uterus (hysterectomy) or ovaries and fallopian tubes (salpingo-oophorectomy) may be available for gender-affirming surgery. Currently, a risk-reducing bilateral salpingo-oophorectomy (BSO) is the most effective means of decreasing ovarian cancer risk in individuals with high-risk hereditary cancer syndromes Guillem et al., 2020). This surgery is performed differently and removes larger amount of tissue than a BSO for non-risk-reducing reasons, such as surgical transition. Surgery to remove the prostate (prostatectomy) is not typically offered as part of a gender-affirming surgery and is also not typically recommended as a risk-reduction option in most cases of high-risk hereditary cancer syndromes. Possible risks associated with gender-affirming surgeries are those of typical surgery-related complications (Coleman et al., 2012). Because there is limited data on the effects that these therapies have on cancer risk, transgender and gender nonconforming patients who present for cancer risk assessment require a more personalized approach to discussing gender-affirming interventions and management options (Sutherland et al., 2020).

1.4 Health Disparities in Cancer Care

There are significant barriers that limit access to gender-affirming therapies and general healthcare services among gender-diverse populations (Berro, et al., 2019; Harb et al., 2019; Haviland et al., 2020; Sacca et al., 2019; Sutherland et al., 2020; Zayhowski et al., 2019). These barriers include lack of competent care, inhibited access to health

insurance, and limited availability of inclusive and comfortable healthcare environments (Edmiston et al., 2016; Harb et al., 2019; Haviland et al., 2020). It has been shown that healthcare providers with professional education and formal training regarding care for lesbian, gay, bisexual, transgender, and questioning (LGBTQ) individuals demonstrate an increase in knowledge and competency when serving these patient populations (Hardacker et al., 2014; Johnson et al., 2016). Patients who are currently employed, have higher annual incomes, have completed higher levels of education, and have health insurance are more likely to adhere to cancer screening and prevention recommendations than those who do not (Johnson et al., 2016). Adults who are of ethnic minorities are more likely than white adults to identify as a gender minority, which can compound these barriers and further limit and prevent access to medical care, ultimately leading to worse health outcomes (Center for Disease Control and Prevention 2018).

Without accurate, standardized documentation of gender diversity in most medical record systems, cancer rates among transgender people are poorly understood. The SEER database does not collect gender identity information, therefore there is no way to determine cancer rates among transgender patients at a population level. Individuals who are at high risk for developing cancer, including individuals with hereditary cancer syndromes, have specific screening recommendations that they should follow based on the specific body tissues they have. Based on the information that is available, transgender individuals have been reported to have extremely low uptake on all cancer screenings. Transgender and gender-nonconforming individuals have lower proportions of routine colorectal cancer screening compared to cisgender individuals (Tabaac et al., 2018). Transgender individuals are also less likely to adhere to breast cancer screening

recommendations (Bazzi et al., 2015). Receiving less cancer surveillance contributes to increased time to cancer diagnosis which leads to increased rates of morbidity and mortality among transgender patients (Sutherland et al., 2020).

High-risk individuals who have at-risk body tissues, regardless of gender, should follow the NCCN Guidelines for hereditary cancer management. However, there are no evidence-based cancer screening guidelines specifically for transgender patients. Breast tissue, one or both ovaries, a uterus, and/or a prostate are of specific importance to transgender individuals, because these body systems are at an increased risk for tumor development with many genetic predisposition syndromes. Additionally, some individuals may have elected to have that tissue removed before the genetic counseling session or may want to learn more about their options for removal of certain at-risk tissues during the session. Transgender men may elect to have chest surgery to create a more masculine appearance, however, this does not eliminate their breast cancer risk, as there is some residual tissue left over (Alaofi et al., 2018; Griepsma et al., 2014).

Some research has been published regarding care for sexual minorities, however, gender diverse individuals are vastly underrepresented in literature regarding cancer screening and genetic counseling (Haviland et al., 2020). The transgender population is generally absent from the little data that does exist on LGBTQ individuals (VandenLangenberg et al., 2011). In the studies that include lesbian, gay, and bisexual patients, genetic counselors report overall that they do not adjust their counseling approaches (Glessner et al., 2012). When it comes to pedigree drawing, genetic counselors have standard nomenclature dictated by NSGC and the NCCN on how to represent different individuals, relationships, and disease status, which, in a cancer

setting, would be cancer diagnoses. The information that exists on how to represent transgender and gender-nonconforming individuals is limited and inconsistent (Berro et al., 2019; Sheehan et al., 2020). The inconsistencies in recommendations from national organizations make it difficult to properly represent these individuals in a standardized, informative, and respectful way.

1.5 Rationale

Little research has been conducted to assess the feelings and experiences of transgender patients regarding current genetic counseling practices. Most of the information that is available comes from case studies or from the perspective of genetic counselors rather than of transgender community members. Additionally, each of these studies calls for a greater exploration of the views of transgender patients themselves. To our knowledge, no research has been conducted to assess the perspectives of transgender individuals who have not had cancer genetic counseling specifically regarding anticipated needs and perceived barriers to accessing genetics care.

The identification of a pathogenic genetic variant that increases someone's risk to develop cancer could impact a transgender patient's decisions about hormonal or surgical medical interventions. There is limited information available on the unique needs of transgender individuals in a cancer genetic counseling session. This population may be a significant portion of the patients seen in clinics, and there are currently no standardized guidelines or recommendations for how to care for transgender patients in a cancer genetics setting (Barnes et al., 2019; Rafferty 2018; Sheehan et al., 2020). To meet the needs of this patient population, patient perspectives must be assessed. This study aimed to determine what aspects of current genetic counseling practice and training need to be

adapted to benefit this patient population. We anticipated that the results of the study would aid in the optimization and standardization of cancer genetic counseling strategies for transgender patients.

1.6 Purpose

This project was conducted to assess the anticipated needs of transgender adults during a cancer genetic counseling session. The primary goal of this study was to determine the informational and psychosocial needs of transgender patients who may present for cancer genetic counseling. This study also aimed to identify motivations and barriers to seeking genetic counseling. The information collected was be compared to previously reported perspectives of cancer genetic counselors on their experiences with counseling transgender individuals about hereditary cancer risk. Finally, through this study, we hoped to recognize themes to assist in standardization of practice and optimization of care of gender minority patients.

This study aimed to provide genetic counseling research surrounding transgender healthcare, which is currently sparse and limited. While there is literature that assesses the needs of patients who identify as a sexual minority, research regarding how genetic counselors can meet the needs of transgender patients specifically is minimal. In the NSGC 2020 Professional Status Survey–an internally administered survey that assesses metrics of current genetic counselors such as demographics, salary, and workforce satisfaction–only two genetic counselors indicated non-binary or third gender identities. The literature that does exist regarding transgender patients is predominantly from the perspective of genetic counselors, most of whom do not identify as transgender or non-binary, leaving the transgender voice largely absent. The informational, psychosocial, and

medical management needs of transgender individuals is likely different from those of cisgender patients. Therefore, it is important to explore the perspectives of these patients to gain information about how best to care for them. Without intentional surveying of gender minority patients, genetic counselors will remain underprepared to care for a portion of the patient population. In existing literature, genetic counselors indicated a desire for more data and education regarding counseling transgender patients (Zayhowski et al., 2019).

Identifying the needs of transgender patients adds to the cultural competence that genetic counselors must have to effectively serve patients, making the information gained from exploring these perspectives valuable to the profession. Cultural competency is a core value of the genetic counseling field, as it encourages genetic counselors to understand each patient's family, community, and culture to better empathize with patients' thoughts, feelings, and values. Understanding the motivations and needs of transgender people who may present for cancer genetic counseling is the only way to competently counsel these patients. It is the duty of genetic counselors to be knowledgeable and sensitive to diverse languages, cultures, beliefs, and backgrounds to provide optimal care (Warren & Wilson 2013). The information gained from surveying patients directly will prepare current and future genetic counselors for interactions with all patients, including transgender and gender nonconforming patients.

We predicted that transgender individuals have needs within the cancer genetic counseling session that are similar to cisgender patients as well as needs that are unique to their gender identity. We expected that transgender individuals would find value in discussing how future healthcare plans regarding gender-affirming therapies could be

impacted by a hereditary predisposition to cancer. Results from this study may also provide clarification on current practices regarding official pedigree standards.

CHAPTER 2

ASSESSING THE ANTICIPATED NEEDS OF TRANSGENDER PATIENTS IN CANCER GENETIC COUNSELING[1]

[1]Baquet, J., Koeller, D., Nunley, P., and Tomlin, K. To be submitted to *Journal of Genetic Counseling.*

2.1 Abstract

Most cancers are sporadic, but 5-10% of all cancer is hereditary, or caused by a heritable genetic mutation. A patient's medical history, family history, genetic test results, intact organs (e.g., ovaries) at an increased risk for developing cancer, and the availability and accessibility of interventions are used to make recommendations for cancer-risk management. Transgender patients have healthcare needs that differ from those of cisgender patients such as expert care related to using hormones or having gender-affirming surgery, as well as unique mental health concerns. Transgender individuals may also experience a greater number of barriers to accessing care than cisgender individuals.

The purpose of this study was to explore the motivations and needs of transgender individuals who may seek cancer genetic counseling. We aimed to determine where current practices could be improved to increase comfortability and inclusivity of transgender patients. Eighty-seven transgender individuals participated in an online questionnaire that asked about their personal perspectives on comfort and preferences regarding current genetic counseling practices.

Most participants reported that they would feel comfortable sharing their pronouns, hormone therapies, and surgical history on an intake form before their genetic counseling appointment. The results suggested that comfort levels between the different current practices regarding pedigree nomenclature had no statistical differences, although most participants would not be comfortable being represented as their sex assigned at birth on a pedigree. When assessing motivations, survey responses demonstrated that most participants would want to discuss how hormone and surgical therapies impact personal

cancer risk. These findings reinforce recommendations from existing literature regarding the adaptation and evolution of current practices to meet the needs of transgender patients while highlighting the need for standardized training in order to provide comprehensive, inclusive care for all patients, regardless of gender identity.

2.2 Introduction

A person's sexuality and gender are made up of many features including gender identity, gender expression, sex assigned at birth, and sexual orientation (see Table 2.1 for a glossary of terms used in this paper). In the United States, where children are typically assigned a sex at birth before they develop their gender identity, over one million Americans identify as transgender and/or nonbinary (Puechl et al., 2019, Rafferty 2018). When a person's gender does not align with their sex assigned at birth, they may choose to express their gender through social changes, hormonal therapies, and/or surgical transitions.

Table 2.1 Glossary of Terms

Term	Definition
Cis/Cisgender	Used to describe a person whose gender identity and gender expression align with sex assigned at birth and culturally defined gender roles.
Gender	Refers to the attitudes, feelings, and behaviors that a given culture associates with a person's assigned sex.
Gender Dysphoria	Discomfort of distress related to incongruence between a person's gender identity, sex assigned at birth, and/or primary sex characteristics.
Gender Expression	The presentation of an individual, including physical appearance, clothing choice and accessories, and behaviors that express aspects of gender identity or role.
Gender Identity	A person's inherent sense of self as it relates to gender.

Nonbinary	Used to describe a person whose gender identity does not align with a binary understanding of gender such as a person who may identify as a combination of genders, who does not define themselves as gendered, or who embodies a third gender. *Nonbinary can include agender, bigender, demigender, genderfluid, gender-nonconforming, gender-neutral, genderqueer, pangender, or traditional third-genders such as Two-Spirits and Muxes.*
Gender Role	The culturally defined pattern of behavior, personality traits, and attitudes that define masculinity and femininity.
Intersex	Used to describe a person who was born with variation in their sex traits and reproductive anatomy such as gonads, internal reproductive organs, external genitalia, and hormone production. *Intersex is not used to describe individuals based on sex chromosome configuration alone.*
LGBTQ	An acronym for "Lesbian, Gay, Bisexual, Transgender, Queer or Questioning, Intersex, and Allies."
"Top Surgery"	Surgical transition through breast augmentation mammoplasty or subcutaneous mastectomy with chest contouring.
"Bottom Surgery"	Surgical transition through reproductive and/or genital organ reconstruction or removal.
Risk-Reducing Mastectomy	A prophylactic surgery that involves the removal of breast tissue. A total mastectomy, complete removal of both breasts including nipples, provides greater risk reduction than a subcutaneous, nipple-sparing, mastectomy.
Sex/Sex Assigned at Birth	Refers to physical and biological traits prior-to or following birth such as sex chromosomes, gonads, internal reproductive organs, and external genitalia. Typically described as male (*AMAB*), female (*AFAB*), or intersex (*AIAB* or *UAAB*).
Transgender	Used to describe the full range of people whose gender identity does not conform to the gender role that is culturally associated with their sex assigned at birth.
Transitioning	The psychological, social, hormonal, surgical and legal processes by which some people strive to more closely align their gender identity and gender expression.
Two-Spirit	A pan-Indian term used to describe a Native American or Alaskan Native who identifies with third gender or an

> alternative gender status. Many tribes and cultures have additional specific terms for their nonbinary genders.

The National Cancer Institute (NCI) has been publicly documenting cancer incidence data online in the Surveillance, Epidemiology, and End Results (SEER) Program since 1973. According to SEER, there were 1,806,590 new cases of cancer in 2020. While most cancers are sporadic, approximately 5-10% of all cancer is hereditary, or caused by a heritable genetic mutation in a cancer predisposition gene (Dalton et al., 2014). When an individual has a genetic predisposition to cancer, they are at an increased risk for developing certain types of cancer, depending on which gene has a mutation. Hereditary cancers tend to occur at younger ages than sporadic cancers and individuals with a hereditary cancer syndrome can have multiple cancers in their lifetime.

Based on these risks, the National Comprehensive Cancer Network (NCCN) has published guidelines for genetic testing for hereditary cancer syndromes and cancer risk management in individuals who have genetic cancer predispositions. These recommendations for cancer screenings and preventative therapies aim to reduce the probability that cancer will occur or reoccur in these individuals. If someone is at an increased risk for developing cancer, recommendations may include earlier and increased cancer surveillance or prophylactic surgical prevention.

Genetic counselors are healthcare professionals with training in both medical genetics and psychosocial counseling. A cancer genetic counselor specializes in evaluating patients for hereditary cancer syndromes. In a typical cancer genetic counseling session, the genetic counselor elicits a patient's personal and family medical history and conducts a risk assessment to determine the likelihood that a patient's cancer or the cancer in their

family may be caused by a genetic predisposition syndrome. The genetic counselor then communicates that risk assessment to the patient, facilitates decision making regarding options for genetic testing, educates the patient on cancer surveillance and prevention, and arranges genetic testing when appropriate (Schneider, 2011). During a genetic counseling session, the counselor also explores psychosocial issues regarding motivations for testing, feelings about the risk information, and overall emotional state of the patient.

When eliciting a patient's family history, the genetic counselor will document the information as a pedigree, or a diagram of the family tree. The National Society of Genetic Counselors (NSGC) has established Practice Guidelines for Human Pedigree Nomenclature, most recently revised in 2008. The NCCN has also published recommendations for pedigree nomenclature that differ slightly from the NSGC Practice Guidelines. The purpose of these guidelines is to create a universally accepted way to accurately and appropriately represent individuals, their relationships, and their medical status for risk assessment and future reference for other medical providers (Bennett et al., 2008). Traditionally, based on the binarization of gender in most Western cultures, females and males have been represented by circles and squares, respectively. If an individual's gender is unknown, they may be represented by a diamond according to the NSGC guidelines or a combination of a square and circle per the NCCN (Bennett et al., 2008; Sutherland et al., 2020). The lack of consensus creates confusion between medical professionals and does not validate the gender identity of transgender patients. There have been suggestions to create a unique symbol for transgender and nonbinary patients within the field of genetic counseling, which underlines the need for standardization and reform of the current practice guidelines (Sheehan et al., 2020; Tuite et al., 2020).

The World Professional Association for Transgender Health (WPATH) published Standards of Care for the Health of Transsexual, Transgender, and Gender-Nonconforming People. These Standards of Care guidelines describe therapies available to aid in the transition from sex assigned at birth to true gender, including hormonal and surgical options. There are some possible risks to taking these hormones, including a potential increase in cancer risk, although much of the data is minimal and sometimes conflicting (Sutherland et al., 2020).

Breast and chest reconstruction may be referred to as "top surgery" and typically involves either a breast augmentation mammoplasty or a subcutaneous mastectomy with chest contouring (Coleman et al., 2012). Subcutaneous, or nipple-sparing mastectomies, are not typically prophylactic bilateral total mastectomies, which is the recommended surgical prevention for some high-risk hereditary breast cancer syndromes (Alaofi et al., 2018; Griepsma et al., 2014; Guillem et al., 2020). Additional surgical transitions may include "bottom surgery," or surgery involving reproductive and/or genital organ reconstruction or removal (Coleman et al., 2012). Surgery to remove reproductive organs such as the uterus (hysterectomy) or ovaries and fallopian tubes (salpingo-oophorectomy) may be available for gender-affirming surgery. Currently, a risk-reducing salpingo-oophorectomy (RRSO) is the most effective means of decreasing ovarian cancer risk in individuals with high-risk hereditary cancer syndromes (Guile et al., 2020). Surgery to remove the prostate (prostatectomy) is not frequently offered as part of a gender-affirming surgery and is also not typically recommended in most cases of high-risk hereditary cancer syndromes. Because there is limited data on the effects that these therapies have on cancer risk, transgender and gender nonconforming patients who

present for cancer risk assessment require a more personalized approach to discussing gender-affirming interventions and management options (Sutherland et al., 2020).

There are significant barriers that limit access to gender-affirming therapies and general healthcare services among gender-diverse populations (Berro, et al., 2019; Harb et al., 2019; Haviland et al., 2020; Sacca et al., 2019; Sutherland et al., 2020; Zayhowski et al., 2019). These barriers include lack of competent care, inhibited access to health insurance, and limited availability of inclusive and comfortable healthcare environments (Edmiston et al., 2016; Harb et al., 2019; Haviland et al., 2020). It has been shown that healthcare providers with professional education and formal training regarding care for lesbian, gay, bisexual, transgender, and questioning (LGBTQ) individuals demonstrate an increase in knowledge and competency when serving these patient populations (Hardacker et al., 2014; Johnson et al., 2016). Patients who are currently employed, have higher annual incomes, have completed higher levels of education, and have health insurance are more likely to adhere to cancer screening and prevention recommendations than those who do not (Johnson et al., 2016). Adults who are of ethnic minorities are more likely than white adults to identify as a gender minority, which can compound these barriers and further limit and prevent access to medical care, ultimately leading to worse health outcomes (Center for Disease Control and Prevention 2018).

Without the ability to document gender diversity in most medical record systems, cancer rates among transgender people are poorly understood. The SEER database does not collect gender identity information, therefore there is no way to determine cancer rates among transgender patients at a population level. Individuals who are at high risk for developing cancer, including individuals with hereditary cancer syndromes, have

specific screening recommendations that they should follow based on what body tissues they have. Breast tissue, one or both ovaries, a uterus, and/or a prostate are of specific importance to transgender individuals, because these body systems are at an increased risk for tumor development with many genetic predisposition syndromes. Based on the information that is available, transgender individuals have been reported to have extremely low uptake on all cancer screenings. Transgender and gender-nonconforming individuals have lower proportions of routine colorectal cancer screening and are less likely to adhere to breast cancer screening recommendations compared to cisgender patients (Bazzi et al., 2015; Tabaac et al., 2018). Receiving less cancer surveillance contributes to increased time to cancer diagnosis which leads to increased rates of morbidity and mortality among transgender patients (Sutherland et al., 2020).

Little research has been conducted to assess the feelings and experiences of transgender patients regarding current genetic counseling practices. In the NSGC 2020 Professional Status Survey–an internally administered survey that assesses metrics of current genetic counselors such as demographics, salary, and workforce satisfaction–only two genetic counselors indicated non-binary or third gender identities. The literature that does exist regarding transgender patients is predominantly from the perspective of genetic counselors, most of whom do not identify as transgender or non-binary, leaving the transgender voice largely absent. Additionally, each of these studies calls for a greater exploration of the views of transgender patients themselves. To our knowledge, no research has been conducted to assess the perspectives of transgender individuals who have not had cancer genetic counseling specifically regarding anticipated needs and perceived barriers to accessing genetics care.

This population may be a significant portion of the patients seen in clinics, and there are currently no standardized guidelines or recommendations for how to care for transgender patients in a cancer genetics setting (Barnes et al., 2019; Rafferty 2018; Sheehan et al., 2020). To meet the needs of this patient population, patient perspectives must be assessed. This study aims to determine what aspects of current genetic counseling practice and training need to be adapted in order to optimize cancer genetic counseling strategies for this patient population. The primary goal of this study was to determine the informational and psychosocial needs of adult transgender patients who may present for cancer genetic counseling while identifying motivations and barriers to seeking genetic counseling.

This study aimed to provide genetic counseling research surrounding transgender healthcare, which is currently sparse and limited. While there is literature that assesses the needs of patients who identify as a sexual minority, research regarding how genetic counselors can meet the needs of transgender patients specifically is minimal (VandenLangenberg et al., 2011). In the studies that include lesbian, gay, and bisexual patients, genetic counselors report overall that they do not adjust their counseling approaches (Glessner et al., 2012). Without intentional surveying of gender minority patients, genetic counselors will remain underprepared to care for a portion of the patient population. In existing literature, genetic counselors indicated a desire for more data and education regarding counseling transgender patients (Zayhowski et al., 2019).

Identifying the needs of transgender patients adds to the cultural competence that genetic counselors must have to effectively serve patients, making the information gained from exploring these perspectives valuable to the profession. Cultural competency is a

core value of the genetic counseling field, as it encourages genetic counselors to understand each patient's family, community, and culture to better empathize with patients' thoughts, feelings, and values. Understanding the motivations and needs of transgender people who may present for cancer genetic counseling is the only way to competently counsel these patients. It is the duty of genetic counselors to be knowledgeable and sensitive to diverse languages, cultures, beliefs, and backgrounds to provide optimal care (Warren & Wilson, 2013). The information gained from surveying patients directly will prepare current and future genetic counselors for interactions with all patients, including transgender and gender nonconforming patients.

We predicted that transgender individuals have needs within the cancer genetic counseling session that are similar to cisgender patients, as well as needs that are unique to their gender identity. We expected that transgender individuals would find value in discussing how future healthcare plans regarding gender-affirming therapies could be impacted by a hereditary predisposition to cancer. Results from this study may also provide clarification on current practices regarding official pedigree standards.

2.3 Materials and Methods

2.3.1 Participants and Recruitment

Participants included English-speaking individuals over the age of 18 who identify as transgender and/or nonbinary. Participants who were not over the age of 18 or did not identify as transgender were excluded from the study. Only English-speaking participants were included in this study due to limited resources for translation and interpretation from English to other languages. Survey exclusion criteria is detailed in Figure 2.1.

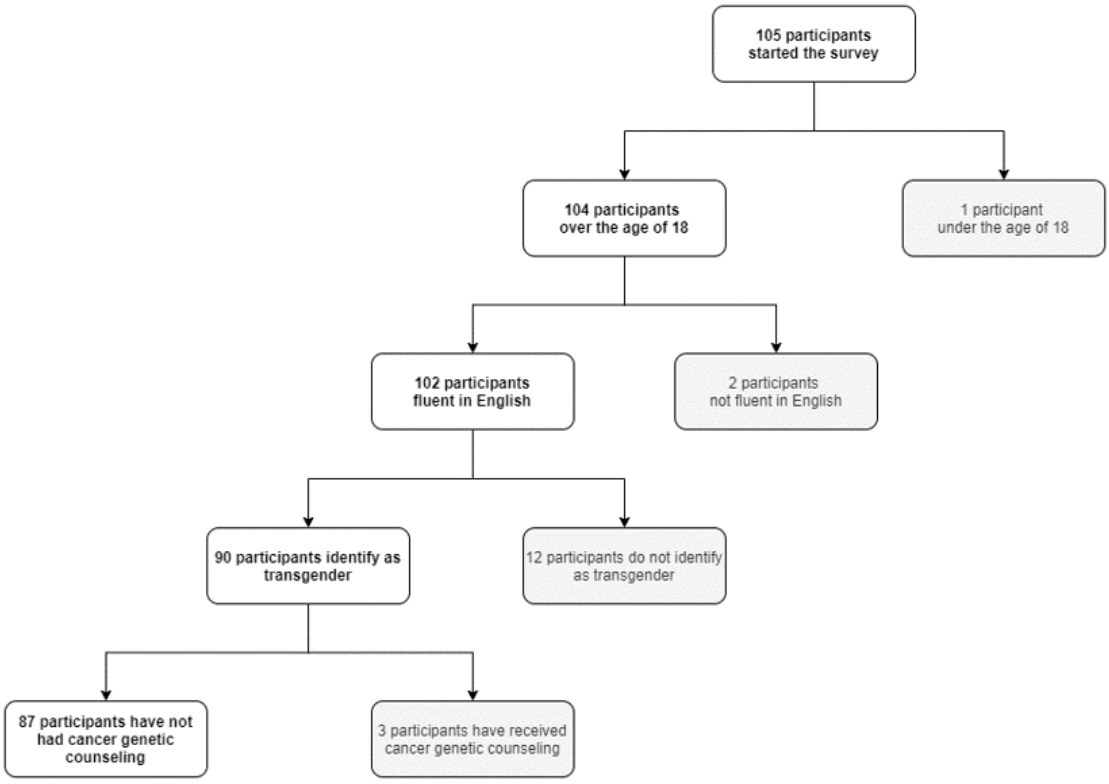

Figure 2.1 Methods by which participant eligibility was filtered to assess for eligibility. Participants included in the grey boxes were excluded from the study and not assessed in data analysis due to unmet inclusion criteria.

Participants were recruited via an advertisement (Appendix A) on multiple Facebook pages for transgender individuals and hereditary cancer risk support groups. The advertisement included a description of the study and a link to the online questionnaire. Participation was voluntary and upon completion of the survey, respondents were given the opportunity to enter a raffle for one of five $25 Visa gift cards. Participants also had the opportunity to express interest in being contacted for a semi-structured phone interview.

2.3.2 Procedure

The questionnaire was administered online through Qualtrics.com and included Likert scale, open-text entry, and multiple choice items. Data regarding gender identity, demographics, motivations for attending a cancer genetic counseling session, perspectives on current practices, personal medical history, and family history of cancer was collected. Participants were able to skip any question or leave the questionnaire at any time, so the completion rate varied. The semi-structured phone interviews were conducted by the principal investigator (JB) and included similar themes as the questionnaire.

Responses were collected from July to October 2020. Of the 105 individuals who began the questionnaire, 87 participants submitted responses that were eligible for data analysis. Eligible responses were reviewed and of the 87 respondents, 61 participants provided answers to the entire questionnaire. Ten participants input their phone numbers into the online software and were contacted for a phone interview. Verbal consent for participation was requested at the beginning of each interview (Appendix C). On average, the interviews lasted 25 minutes (range 15-40 minutes). Interviews were recorded on the PI's password protected computer and transcribed verbatim.

2.3.3 Data Analysis

Quantitative data were analyzed using descriptive statistics, reliability, and correlations as appropriate. Quantitative data were analyzed using Microsoft Excel software to perform Kruskal-Wallis (KW) test and Mann-Whitney U test for differences and associations between groups. Figures and tables were constructed using Microsoft PowerPoint software. The level of comfort and agreeability to several statements was

assessed using a Likert scale (1=strongly disagree; 5=strongly agree) and were represented with descriptive statistics (percentages, frequencies, and means). Due to lack of responses in the open-text entry fields, the qualitative data were descriptively summarized. Quotations from open-text entries and interviews were extracted and organized into categories when available.

2.4 Results

2.4.1 Demographic Information

Of the 87 eligible respondents, 100% disclosed their gender identity with most participants identifying with more than one gender (Table 2.2). Demographic information such as age, race and ethnicity, education and employment, etc. was provided for 61 participants (Table 2.3). The majority of participants were between 18- and 38-years-old, with most participants being 25- to 31-years-old. Almost all participants identified as White and Non-Hispanic. Participant education level ranged from some high school education to completed graduate degrees. Employed respondents with private health insurance were more likely to receive care from a center that specializes in transgender healthcare than participants who were unemployed or uninsured (Figure 2.2).

Table 2.2 Participant Gender Identity Information

Gender	Total (N)	Percent (%)
Agender	5	8%
Bigender	2	3%
Demigender	2	3%
Genderfluid	5	8%
Genderqueer	9	15%
Gender-nonconforming	8	13%
Gender-neutral	3	5%

Gender-questioning	1	2%
Gender-transitioning	4	7%
Nonbinary	20	33%
Transgender man	27	44%
Transgender masculine	11	18%
Transgender feminine	9	15%
Transgender woman	17	28%
Two-spirit	2	3%
Prefer to self-describe	3	5%
Prefer not to say	0	0%

Table 2.3 Participant Demographic Information

Participant characteristic	Total (N)	Percent (%)
Age		
18-24 years old	15	25%
25-31 years old	18	30%
32-38 years old	12	20%
39-45 years old	8	13%
46-52 years old	1	2%
53-59 years old	5	8%
60-66 years old	1	2%
67-73 years old	1	2%
74 years or older	0	0%
Prefer not to say	0	0%
Race		
African American or Black	0	0%
American Indian or Alaskan Native	1	2%
Asian	2	3%
Caucasian or White	56	92%
Pacific Islander	0	0%
Prefer to self-describe	0	0%

Prefer not to say	2	3%
Ethnicity		
Hispanic or Latinx	5	8%
Not Hispanic or Latinx	53	87%
Prefer not to say	3	5%
Education level		0%
Some high school	3	5%
High school diploma or equivalent	3	5%
Some college	22	36%
Technical, trade, or vocational training	2	3%
College graduate	17	28%
Graduate degree	12	20%
Prefer not to say	2	3%
Employment		
Student	10	16%
Homemaker	1	2%
Military	0	0%
Employed	39	64%
Unemployed	5	8%
Retired	1	2%
Unable to work	3	5%
Prefer not to say	2	3%
Income		
$29,999 or less	16	26%
$30,000-39,999	11	18%
$40,000-49,999	8	13%
$50,000-59,999	4	7%
$60,000-69,999	7	11%
$70,000 or more	13	21%
Prefer not to say	2	3%
Insurance		

No health insurance	9	15%
Private health insurance	40	66%
Public health insurance	10	16%
Prefer not to say	2	3%
Receiving healthcare from a center that specializes in transgender healthcare		
Yes	37	61%
No	22	36%
Prefer not to say	2	3%

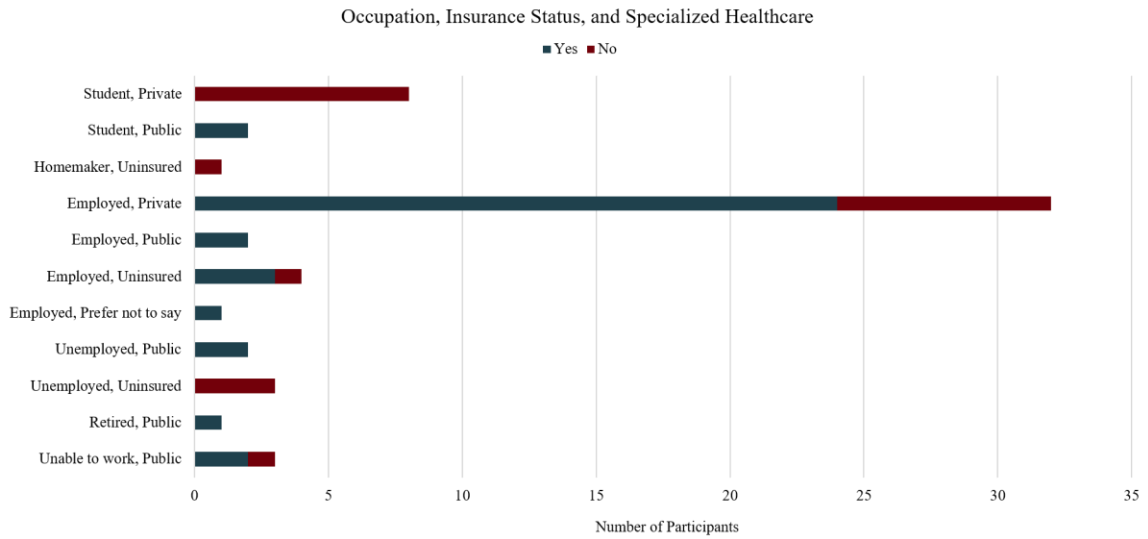

Figure 2.2 The impact of employment and health insurance status on receiving specialized healthcare at a center for transgender patients.

2.4.2 Personal and Family History of Cancer

Four participants had a previous diagnosis of cancer including breast cancer at 45-years-old, cervical cancer at 25-years-old, bladder cancer at 29-years-old, and laryngeal cancer at 69-years-old. Although approximately half (42%) respondents reported limited knowledge of their family health history, 63 participants indicated a family history of at least one first- or second-degree relative who was diagnosed with cancer (Figure 2.3). The total number of cancer diagnoses reported exceeds 228 cases. On average,

participants had about 3 relatives who have been diagnosed with cancer. The most frequently reported types of cancer, when known, for these relatives was breast cancer and melanoma which were present in 43% and 41% of families, respectively.

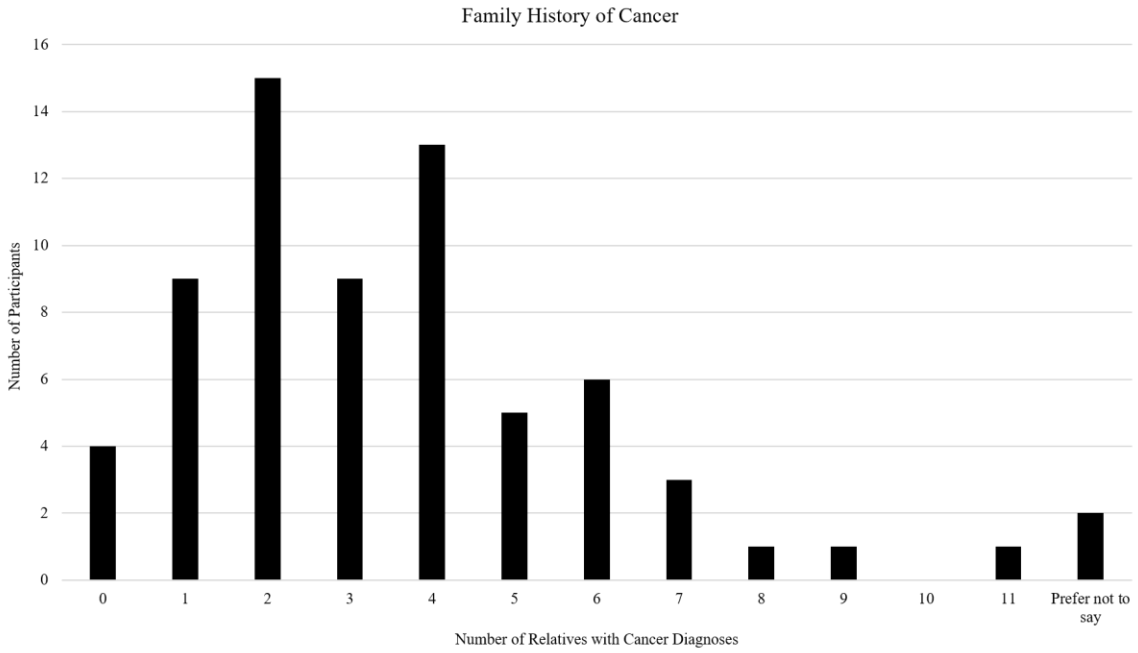

Figure 2.3 Number of participants that indicated how many of their first- and second-degree relatives have been diagnosed with cancer.

2.4.3 Current Practices

The questionnaire asked participants to rate their level of comfort with different statements addressing the current practices of cancer genetic counselors using a Likert scale (1=strongly disagree; 4=strongly agree). Participants were asked generally about the importance of inclusivity within healthcare offices and appointments, preferred methods of disclosing information, and specifically about comfort communicating with family members (Table 2.4; Table 2.5).

Most participants (89%) indicated that it is important to have visible signs in the waiting room or lobby regarding LGBTQ inclusivity. A large portion of participants

agreed that it is important for medical professionals use appropriate and inclusive language both on intake forms and during the appointment. Participants generally indicated that they would feel comfortable sharing cancer risk information with their relatives, both with and without assistance from a genetic counselor (Table 2.4; Figure 2.4). Most participants (79%) who indicated they would not share cancer risk information with their families without the assistance of a genetic counselor said they would if they did have that assistance in the form of a family letter, phone call, or family appointment. Some participants indicated that the importance of each item would depend on the specific situation or circumstances and elaborated in the open-text entry field associated with that section of the survey.

Table 2.4 Comfort Levels Sharing Information Related to Cancer Risk

Statement	Strongly disagree N (%)	Disagree N (%)	Agree N (%)	Strongly agree N (%)	It depends N (%)
If I was at an increased risk to develop cancer due to a genetic cause, I would feel comfortable sharing this with my relatives without assistance from a genetic counselor.	4 (6.4%)	10 (15.9%)	21 (33.3%)	17 (27%)	10 (15.9%)
If I was at an increased risk to develop cancer due to a genetic cause, I would feel comfortable sharing this with my relatives with assistance from a	0 (0%)	5 (8.1%)	32 (51.6%)	16 (25.8%)	7 (11.3%)

genetic counselor like a family letter, phone call, or follow-up appointment.				

Table 2.5 Comfort Levels Disclosing Information Related to Gender

Statement	Strongly disagree N (%)	Disagree N (%)	Agree N (%)	Strongly agree N (%)
I would prefer being asked directly about my pronouns over voluntarily sharing them.	3 (4.8%)	2 (3.2%)	25 (40.3%)	29 (46.8%)
I would feel comfortable sharing my pronouns on an intake form before my genetic counseling appointment.	0 (0%)	1 (1.6%)	22 (36.1%)	38 (62.3%)
I would feel comfortable sharing my pronouns with my genetic counselor as soon as I am called from the waiting room.	5 (8.2%)	9 (14.8%)	25 (41%)	22 (36.1%)
I would feel comfortable sharing my pronouns with my genetic counselor when I am inside their office.	1 (1.6%)	4 (6.6%)	30 (49.2%)	26 (42.6%)
I would feel comfortable sharing my pronouns after the genetic counselor shared theirs.	1 (1.6%)	3 (4.9%)	19 (31.2%)	37 (60.7%)
I would feel comfortable disclosing any gender-affirming hormones and surgeries I've had on an intake form before my genetic counseling appointment.	0 (0%)	11 (18%)	21 (34.4%)	28 (45.9%)
I would feel comfortable disclosing any gender-affirming hormones and surgeries I've had during my genetic counseling appointment.	0 (0%)	3 (4.9%)	26 (42.6%)	30 (49.2%)

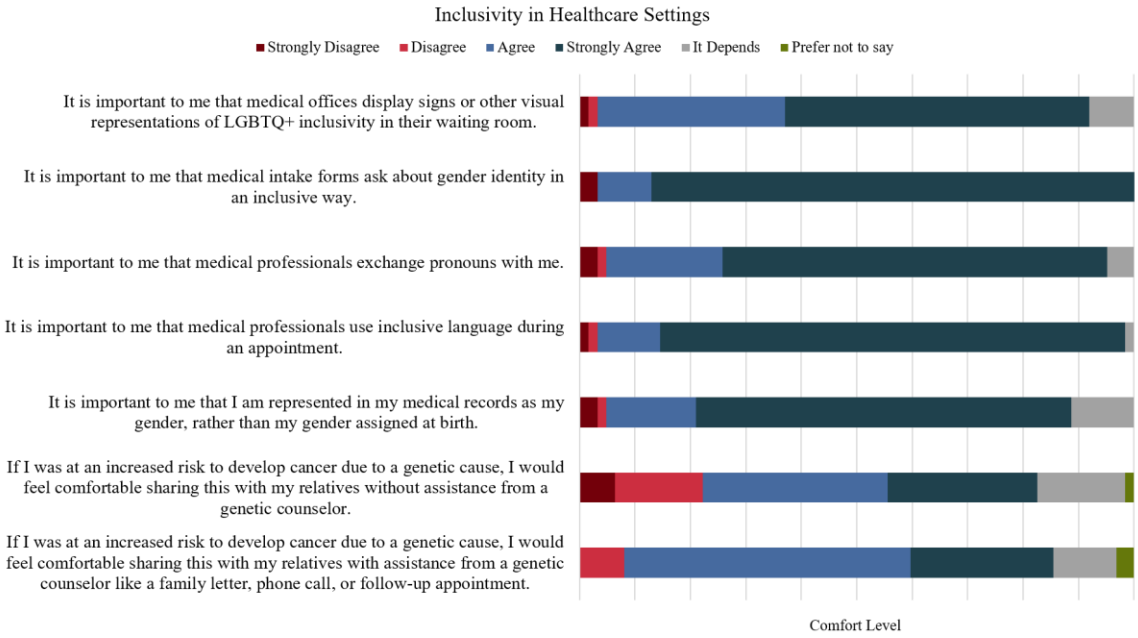

Figure 2.4 Participant responses to questions regarding the importance of inclusivity before and during genetic counseling appointments and comfort with sharing cancer risk information with relatives.

Although comfort levels between sharing pronouns, hormone history, and surgical history was not significantly different between being asked on an intake form or in person, respondents expressed the highest level of agreement to "I would feel comfortable sharing my pronouns on an intake form before my genetic counseling appointment" ($p=0.65$; Figure 2.5). Between available options for pedigree nomenclature, respondents were significantly less comfortable with being represented as their sex assigned at birth ($H(2)=39.88$, $p=2.18E-09$; Figure 2.6).

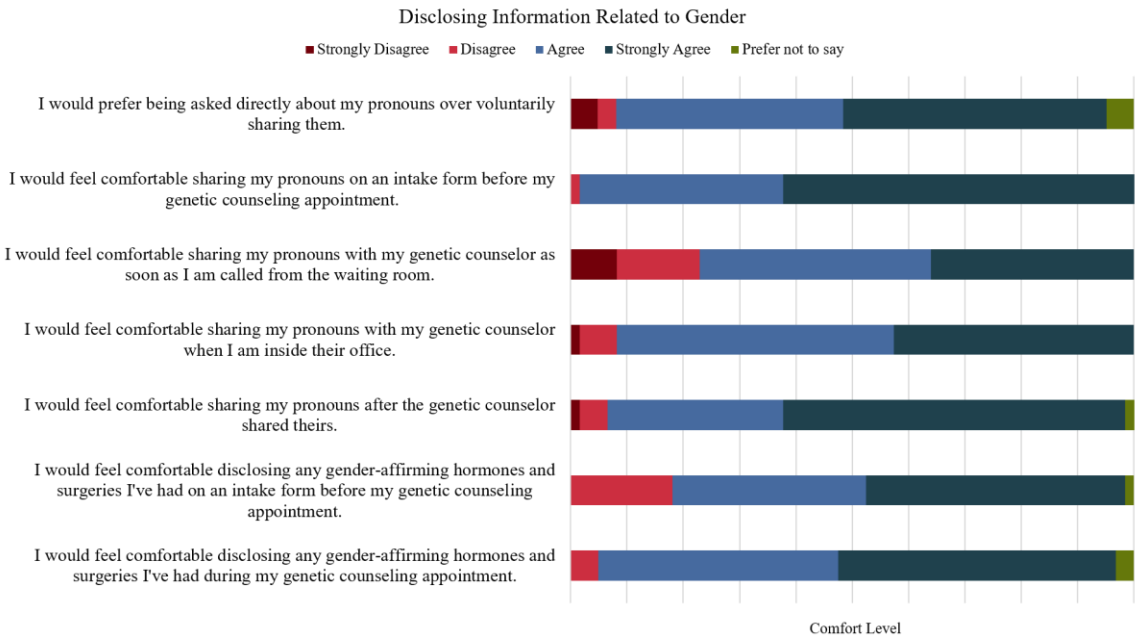

Figure 2.5 Participant responses to questions regarding the most comfortable way to disclose pronouns, hormone use, and surgical history.

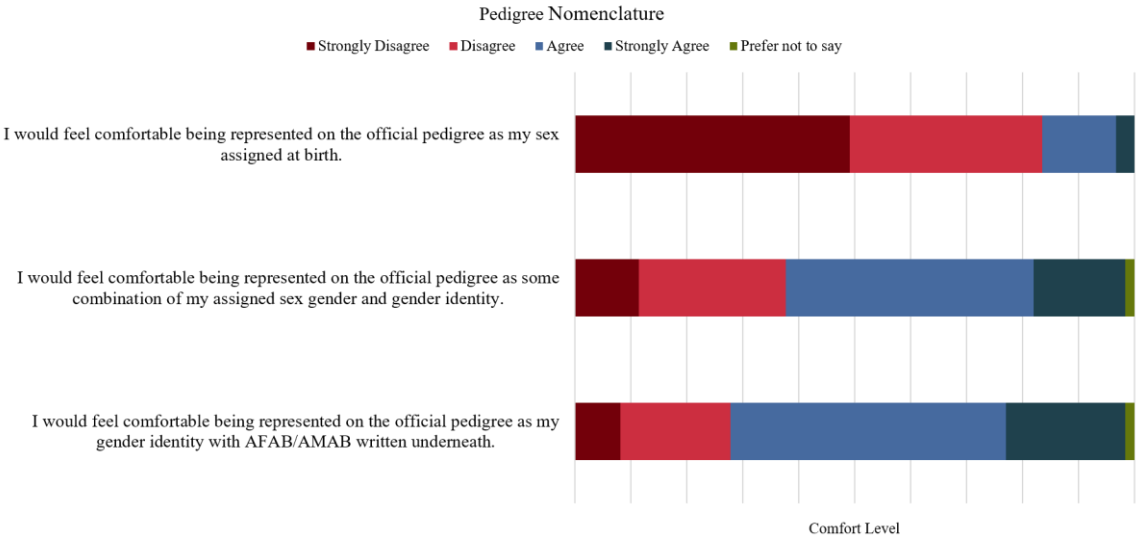

Figure 2.6 Participant responses to questions regarding comfort levels with current pedigree nomenclature.

2.4.4 Motivations

Roughly two-thirds of participants strongly agreed that they would value discussing hormone therapies and gender-affirming surgeries in relation to personal cancer risk during a genetic counseling session. Other motivations for which participants expressed a

high level of agreeance with included to gain information about personal cancer risk assistance as well as understanding options for cancer prevention, risk-reduction, and detection (Table 2.6; Figure 2.7; Figure 2.8). Although a majority of participants expressed agreement with the statements "I would value learning about the cancer risk for my relatives," and "I would value discussing how my cancer risk could impact my family planning decisions," these ranked the lowest among the agreed with statements (Figure 2.7).

Table 2.6 Motivations for Seeking Cancer Genetic Counseling

Statement	Strongly disagree N (%)	Disagree N (%)	Agree N (%)	Strongly agree N (%)
I would value learning about my personal cancer risk.	0 (0%)	0 (0%)	30 (50%)	30 (50%)
I would value learning about the cancer risk for my children.	4 (6.9%)	9 (15.5%)	16 (27.6%)	16 (27.6%)
I would value learning about the cancer risk for my relatives.	1 (1.7%)	7 (11.9%)	32 (54.2%)	19 (32.2%)
I would value having someone to help me understand how my personal and family history impacts my cancer risk.	0 (0%)	1 (1.7%)	29 (49.2%)	29 (49.2%)
I would value discussing my thoughts and feelings about my cancer risk.	0 (0%)	3 (5.1%)	29 (49.2%)	26 (44.1%)
I would value discussing my thoughts and feelings about my genetic test results.	0 (0%)	1 (1.7%)	26 (44.1%)	31 (52.5%)
I would value discussing my thoughts and feelings about cancer screening procedures.	0 (0%)	2 (3.4%)	31 (52.5%)	26 (44.1%)
I would value discussing how my cancer risk could impact my family planning decisions.	2 (3.4%)	10 (17%)	20 (33.9%)	18 (30.5%)

I would value discussing hormone therapies and gender-affirming surgeries as they relate to my cancer risk.	1 (1.7%)	0 (0%)	20 (34.5%)	37 (63.8%)
I would value having someone to help me understand my options for cancer detection.	0 (0%)	2 (3.4%)	27 (45.8%)	30 (50.9%)
I would value having someone to help me understand my options for cancer prevention and risk-reduction.	0 (0%)	0 (0%)	23 (39%)	36 (61%)
I would value having someone to help me decide if genetic testing is right for me.	0 (0%)	0 (0%)	27 (45.8%)	30 (50.9%)
I would value discussing costs relating to genetic testing and follow-up screening.	0 (0%)	1 (1.7%)	23 (39%)	35 (59.3%)
I would value having someone to help me understand the privacy and confidentiality policies surrounding genetic test results.	0 (0%)	3 (5.1%)	29 (49.2%)	26 (44.1%)

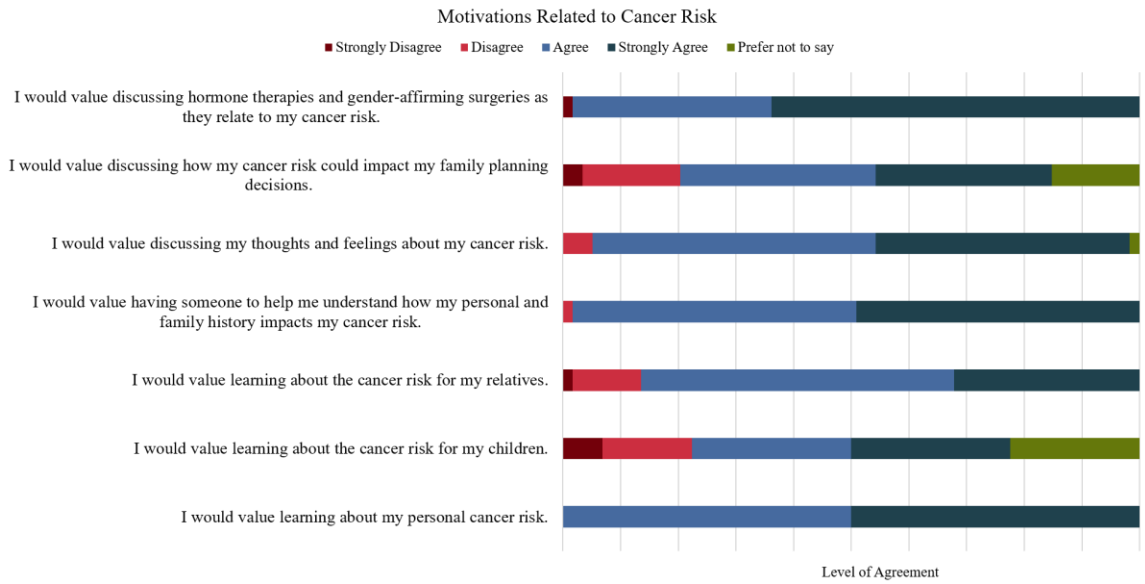

Figure 2.7 Participant responses to questions regarding motivations related to personal and family cancer risk.

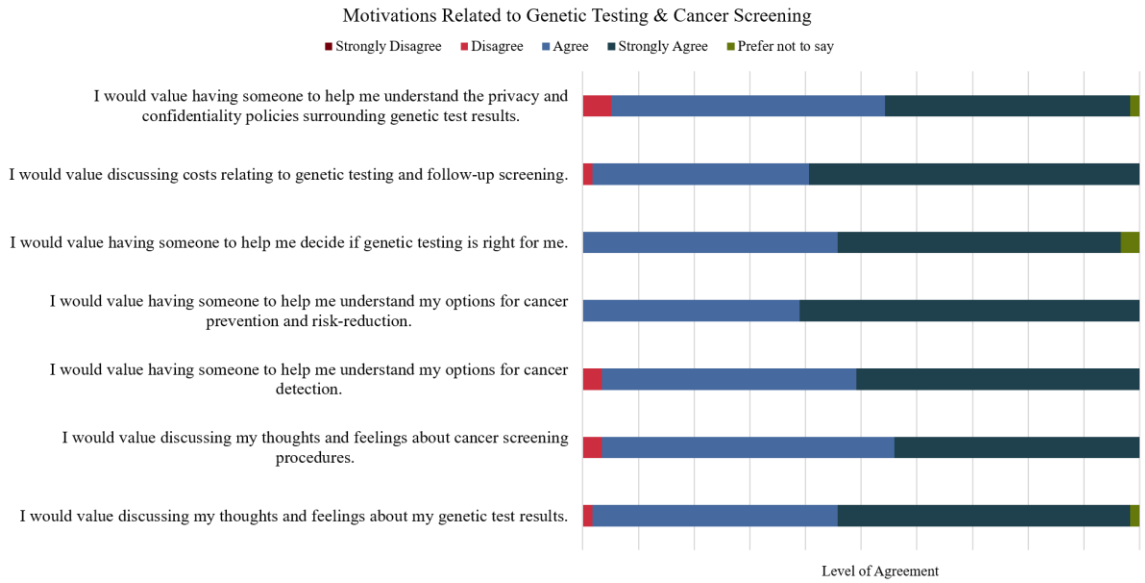

Figure 2.8 Participant responses to questions regarding motivations related to genetic testing and cancer detection, prevention, and risk-reduction.

Most respondents have taken or have considered taking gender-affirming hormones (Table 2.7). A majority of participants have had or have considered having chest or breast surgery. Almost half of participants who were born with ovaries have had or considered having an oophorectomy and over half of respondents who were born with a uterus have had or considered having a hysterectomy. Of the participants who were born with a prostate, most have not had or considered having a prostatectomy.

Table 2.7 Participant Hormone and Surgical History Information

	Yes **N (%)**	**No** **N (%)**	**Prefer not to say** **N (%)**
Have you ever taken, or considered taking, gender-affirming hormones?	53 (88%)	7 (12%)	0 (0%)
Have you ever had, or considered having, surgery on your breast tissue (lumpectomy, mastectomy, breast augmentation, chest reconstruction)?	49 (82%)	8 (13%)	3 (5%)
If you were born with ovaries, have you ever had, or considered having,	19 (48%)	10 (25%)	11 (28%)

surgery to remove your ovaries (oophorectomy)?			
If you were born with a uterus, have you ever had, or considered having, surgery to remove your uterus (hysterectomy)?	22 (54%)	10 (24%)	9 (22%)
If you were born with a prostate, have you ever had, or considered having, surgery to remove your prostate (prostatectomy)?	4 (21%)	14 (74%)	1 (5%)

2.4.5 Qualitative Results

In each survey, participants had the opportunity to answer open-text response questions and provide information or elaborate on the responses they provided. Of the ten individuals who were contacted, six participants completed a follow-up telephone interview. Direct quotes are extracted from open-text entries and telephone interview.

When given the opportunity to expand on their family history of cancer, some individuals specified which relatives had cancer, provided information about "other" types of cancer, and/or explained why they did not know about their family history of cancer. The amount of detail provided by each participant varied greatly and did not correlate with the amount of information the participant indicated they knew about their family medical history. Listed below are quotes from participants regarding their family history of cancer.

1. *"Dad died of prostate cancer. Brother had a prostatectomy."*
2. *"Great-grandmother had breast cancer."*
3. *"Grandmother had colon cancer in her early 50s. At 73 she developed stomach, liver, and other cancer that had metastasized from which she passed of within 2*

weeks of diagnosis. Prostate cancer from maternal grandfather's brother. Bladder cancer from maternal uncle."

4. *"Maternal grandmother and sister both died from ovarian and endometrial cancer."*
5. *"The 'other' cancers are basal cell carcinomas."*
6. *"My aunt had breast cancer and received chemo and they removed her breast tissue on that side. Another aunt had [ovarian] cancer."*
7. *"2 aunts of mother's side, my father, 1+ on my father's side. Family is fractured, data incomplete."*
8. *"I don't know because I'm adopted with no contact with [my biological] family. I was diagnosed with a ductal hyperplasia"*

Listed below are quotes from participants regarding comfort levels with current genetic counseling pedigree nomenclature and suggestions for fostering inclusivity as a healthcare professional.

9. *"I understand in the medical field it can be important to look at birth gender when assessing risks or looking at treatments, so it wouldn't bother me to have that information be a part of the process. I would just prefer being referred to as my chosen gender and pronouns when we're speaking and on any regular paperwork."*
10. *"I usually don't mind sharing any hormone related medication on intake forms, surgeries are usually left unstated unless such information is deemed necessary."*
11. *"I would only be comfortable with the AFAB identifier after gender affirming surgery."*

12. *"We live our lives mostly telling people who we are. It's much nicer to write it down and pass it to said people of an office rather than someone saying out loud in front of strangers. Also, being asked is so much nicer and more comfortable than having to correct someone or tell them straight forward what you prefer."*
13. *"Having a pronoun field on the intake sheet helps to standardize sharing pronouns and is a great step to making the process more inclusive."*
14. *"I've been very fortunate in my medical experiences so far, but I'm always a little nervous when seeing a new doctor for the first time. Seeing some sort of LGBT+ support information in the lobby or on their website would be very comforting."*
15. *"Medical facilities should always be safe places because you are poked and tested so much anyways. It is important to still keep up with doctors to know the health of your body and you should never feel scared to go to the doctors you need [because you are transgender]. [Healthcare is] needed to sustain a long and healthy life. So, it should be apparent from the moment you touch the front door of an office with a sticker or sign saying, "we include everyone including LGBTQ+". Some of the worst moments of my life have been in a doctor's office and I don't want that for others."*

Listed below are quotes from participants regarding potential motivations for seeking cancer genetic counseling. Some participants expanded on what they would like to have addressed during a session and other provided insight on why certain statements were less motivating.

16. *"We always hear that there are risks from taking [hormones] but there is never a long conversation about exactly what those risks are, especially about cancer, nobody really asks about that."*
17. *"Given our understanding of how transgender hormone therapies and surgeries are still not well understood in the long term, I would find it very helpful to have some of that information included in an overview of my potential cancer risks."*
18. *"I have always planned to foster/adopt children and do not plan on having any biological children."*
19. *"It would be nice to have someone say, 'this is your risk and here's why,' so if a genetic counselor could do that, I would go."*
20. *"I feel like I would just want to make sure I was doing the right cancer screenings, so I don't get blindsided by anything."*

2.5 Discussion

2.5.1 Discussion

This study explored perspectives of transgender individuals who may present for hereditary cancer genetic counseling regarding the primary informational and psychosocial needs, motivations for seeking cancer counseling, and barriers to accessing or attending genetic counseling. The purpose of this study was to gain a better understanding of what areas of current genetic counseling practice and formal education need to be evolved to provide optimized, comprehensive, and inclusive care for this patient population. While the perceptions of genetic counselors on trans-inclusive counseling have been investigated previously (Barnes et al., 2019; Berro et al., 2019; Sacca et al., 2019; Sheehan et al., 2020; Zayhowski, et al., 2019) each of these studies

calls for a direct assessment of patient-reported educational and psychosocial needs of transgender patients.

We predicted that transgender and nonbinary individuals would have needs within the cancer genetic counseling session that are similar to cisgender patients, as well as needs that are unique to their gender identity. We expected that participants would find value in and be motivated by the opportunity to discuss how future healthcare plans regarding hormone and surgical transitioning could be impacted by a hereditary predisposition to cancer. We also aimed to obtain clarification on opinions on the current, official pedigree nomenclature standards.

The information revealed in this study is similar to what has been previously reported for transgender patients in genetic counseling in general, emphasizing the importance of adapting to the self-reported needs of this patient population. Specific themes highlighted are that transgender individuals have both similar and diverse motivations and needs related to hereditary cancer genetic counseling, patient agreeance with the current and proposed pedigree nomenclature varies between methodologies, and that there are barriers to accessing care that genetic counselors should be aware of and take into consideration while counseling transgender patients.

Common motivations among both transgender and cisgender individuals regarding seeking hereditary cancer genetic counseling include learning about personal cancer risk, attempting to better understand options for cancer prevention and risk-reduction, and having someone to help that individual decide if, and when, genetic testing may be right for them. Since every respondent either agreed or strongly agreed with those statements, it can be inferred that these motivations are not impacted by gender identity and would be

the same across both cisgender and transgender patient populations. All respondents agreed with the statement about cancer prevention and risk-reduction was unexpected because transgender patients typically have an extremely low uptake and adherence to most cancer screening recommendations compared to cisgender patients of the same age and health status (Bazzi et al., 2015; Tabaac et al. 2018). Patients who are currently employed, have higher annual incomes, have completed higher levels of education, and have health insurance are more likely to adhere to cancer screening and prevention recommendations than those who do not (Johnson et al., 2016). A potential explanation for the strong agreeance with learning about cancer prevention and risk-reduction could be the unequal distribution of participants who are employed and have private health insurance. We found that these participants are more likely to receive medical care from a center that specializes in transgender healthcare, which could increase comfort and access to cancer screenings that transgender patients may not otherwise seek.

Roughly half of participants said they would be motivated to learn about the cancer risk for their children and other relatives. For most cisgender cancer genetic counseling patients, genetic counselors discuss familial implications and how these family members could be impacted by the patient pursuing genetic testing, in terms of both benefits and limitations. Although many participants indicated agreeance with learning this information, the concordance was not as high as that seen for other topics. This may result in less time needing to be spent on this topic in a cancer genetic counseling session. Unsurprisingly, all but one participant agreed or strongly agreed that discussing hormone therapies and gender-affirming surgeries as they relate to personal cancer risk would be a motivating factor for seeking genetic counseling, which is a need specific to this patient

population. Most respondents reported that they have or have considered both surgical and hormonal therapies as a part of their transition process. Participants cited the lack of concrete information regarding cancer risks associated with hormone use as one reason that they would like to discuss hormone use during a genetic counseling session. There are some cisgender patients who may have had previous chest or breast surgery, an oophorectomy, a hysterectomy, or taken hormones, however the frequency of these surgical interventions and hormonal therapies in the cisgender population is significantly less than in the transgender population. In addition, depending on age at time of evaluation, transgender patients may have a longer history of hormone use.

Participants were asked to indicate their level of agreement with the comfortability, and therefore appropriateness, of current guidelines and proposed changes to pedigree nomenclature regarding gender. Most participants disagreed or strongly disagreed with being represented as their sex assigned at birth on a pedigree, which is one interpretation of the latest NSGC guidelines which indicate that it is appropriate to assign gender based on phenotype. This information supports previous data that calls for revision and standardization of pedigree nomenclature to increase comfort of patients while maintaining accuracy (Barnes et al., 2019; Rafferty 2018; Sheehan et al., 2020). Participants were most comfortable being represented as their gender identity with an abbreviation of their sex assigned at birth under the symbol. Genetic counselors and transgender individuals alike have proposed an additional symbol, a hexagon, that would indicate a nonbinary individual rather than not specifying their gender (Sheehan et al., 2020; von Vaupel-Klein, & Walsh, 2020). The data from this study supports that proposal, as most participants would feel most comfortable being represented as their

gender identity, and there are currently no symbols outside of the binary male and female, or unknown gender. It is inappropriate to indicate that a transgender or nonbinary patient is an unknown gender if they identify with either, or both, of those genders.

2.5.2 Practice Implications

Patients expressed that it would be more comfortable to share pronouns and hormonal and surgical interventions on an intake form prior to the genetic counseling appointment, as opposed to in person during the visit. Some individuals expressed that having an inclusive intake form is a way to build trust between the healthcare provider and the patient before they meet. Genetic counselors seeing patients for hereditary cancer evaluations, as well as in other settings, should make a conscious effort to revise their current intake procedures to include the option for "other" genders besides the binary male and female in addition to asking about pronouns, which may go along with "preferred name" or "title" sections that are already a part of many institution's intake paperwork. Increased comfort with providers builds trust between that provider and the patient, which may ultimately lead to stronger adherence to recommendations, such as a cancer screening.

Creation and implementation of a standardized pedigree nomenclature that includes appropriate designations for transgender and nonbinary individuals is a crucial next step for the profession. A revision of the current NSGC Standardized Pedigree Nomenclature will allow this patient population to be represented accurately while setting the example for other professional societies to make adjustments to their pedigree nomenclature that supports inclusive patient care.

Since most patients were motivated by the desire to discuss how their hormone and surgical history impacts their cancer risk, it is important for genetic counselors to be aware of which hormone therapies and surgical approaches are commonly available to transgender patients and how these may be similar or different to hormone therapies and surgeries that may be recommended or included in cancer management of cisgender patients. For example, it would be important for cancer genetic counselors to know gender-affirming masculinizing top surgeries do not reduce breast cancer risk equivalently to a risk-reducing bilateral mastectomy, which is recommended in some cases of hereditary cancer. It has been shown that healthcare providers with professional education and formal training regarding care for lesbian, gay, bisexual, transgender, and questioning individuals demonstrate an increase in knowledge and competency when serving these patient populations (Hardacker et al. 2014; Johnson et al. 2016). Therefore, genetic counselors would benefit from formal education either during their training programs or as a part of continuing education.

2.5.3 Limitations and Future Research

The results of this study are limited by a small sample size (n=87), which inhibited the ability to assess significance of data. Sample size may be partially explained by the requirement that participants be fluent in English reading and writing. In addition, our demographics were heavily skewed towards college-educated, Caucasian participants, therefore these findings may not be generalized to ethnic minority transgender individuals or transgender individuals who have not entered any post-secondary education. The utility of investigation of the same themes in transgender people of racial and ethnic minorities as well as diverse education levels lies in comparing the data from

this study and similar studies to data gathered from a population where additional intersectional health equities are present.

In order to be eligible for participation in this study, participants had to be 18 years of age or older. Our study was primarily composed of respondents between the ages of 18- and 38-years-old. Identified themes from this study may not be representative of other cohorts. It may be beneficial in future research studies to explore the perspectives of transgender individuals who are under the age of 18, as it is more likely that minors have not yet begun hormonal or surgical therapies, which may have an effect on motivations and needs in a cancer genetic counseling setting. This information could inform when transgender patients should be asked about family history of cancer and henceforth referred to genetic counseling when appropriate. Providing early cancer risk education may impact transgender medical management in ways that are not well studied.

CHAPTER 3

CONCLUSIONS

Transgender individuals have both similar and diverse motivations and needs related to hereditary cancer genetic counseling when compared to cisgender individuals. Common motivations among both patient populations include learning about personal cancer risk, attempting to better understand options for cancer prevention and risk-reduction, and having someone facilitating the decision-making process surrounding hereditary cancer genetic testing.

Implications regarding children and other relatives was not found to be strongly motivating, although these are common motivations in cisgender patients. Transgender patients have unique needs in terms of risk education regarding hormone use and surgical interventions. Many transgender individuals will have or will consider both hormonal and surgical interventions, which may or may not impact cancer risk. Genetic counselors should be prepared to address this topic during sessions with transgender patients, which would be best supported by formal education on transgender healthcare needs.

Transgender patients may be more comfortable disclosing information regarding pronouns and hormonal and surgical history on an intake form, rather than in person with the genetic counselor. It is important that genetic counselors take this into consideration when creating and revising intake forms. Allowing patients to indicate a gender other than male or female, or list their pronouns, is one way to be inclusive and foster a comfortable setting for transgender patients. Another effort towards inclusivity would be

the creation and implementation of a standardized pedigree nomenclature that includes appropriate designations for transgender and nonbinary individuals, taking their suggestions and preferences into consideration.

REFERENCES

Alaofi, R. K., Nassif, M. O., & Al-Hajeili, M. R. (2018). Prophylactic mastectomy for the prevention of breast cancer: Review of the literature. *Avicenna Journal of Medicine, 8*(3), 67–77. https://doi:10.4103/ajm.AJM_21_18

Barnes, H., Morris, E., & Austin, J. (2019). Trans-inclusive genetic counseling services: Recommendations from members of the transgender and non-binary community. *Journal of Genetic Counseling*. https://doi:10.1002/jgc4.1187

Bazzi, A. R., Whorms, D. S., King, D. S., & Potter, J. (2015). Adherence to mammography screening guidelines among transgender persons and sexual minority women. *American Journal of Public Health, 105*, 2356–2358. https://doi:10.2105/AJPH.2015.302851

Bennett, R. L., French, K. S., Resta, R. G., & Doyle, D. L. (2008). Standardized human pedigree nomenclature: Update and assessment of the recommendations of the National Society of Genetic Counselors. *Journal of Genetic Counseling*, 17(5), 424–433. https://doi:10.1007/s10897-008-9169-9

Berro, T., Zayhowski, K., Field, T., Channaoui, N., & Sotelo, J. (2019). Genetic counselors' comfort and knowledge of cancer risk assessment for transgender patients. *Journal of Genetic Counseling*. https://doi:10.1002/jgc4.1172

Cella, D., Hughes, C., Peterman, A., Change, C. H., Peshkin, B. N., Schwartz, M. D., Wenzel, L., Lemke, A., Marcus, A. C., & Lerman, C. (2002). A brief assessment of concerns associated with genetic testing for cancer: The multidimensional

impact of cancer risk assessment (MIRCA) questionnaire. *Health Psychology*, 21(6), 564–572 . https://doi:10.1037/0278-6133.21.6.564

Centers for Disease Control and Prevention. Behavioral Risk Factor Surveillance System overview: BRFSS 2018. Atlanta, Georgia: U.S. Department of Health and Human Services, Centers for Disease Control and Prevention; 2019. https://www.cdc.gov/brfss/annual_data/annual_2018.html

Cicero, E. C., Reisner, S. L., Merwin, E. I., Humphreys, J. C., & Silva, S. G. (2020). The health status of transgender and gender nonbinary adults in the United States. PLOS One, 15(2), e0228765. https://doi:10.1371/journal.pone.0228765

Coleman, E., Bockting, W., Botzer, M., Cohen-Kettenis, P., DeCuypere, G., Feldman, J., Fraser, L., Green, J., Knudson, G., Meyer, W.J., Monstrey, S., Adler, R. K., Brown, G. R., Devor, A. H., Ehrbar, E., Ettner, R., Eyler, E., Garofalo, R., Karasix, D. H.,…Zucker, K. (2012). Standards of care for the health of transsexual, transgender, and gender-nonconforming people, Version 7. *International Journal of Transgenderism, 13(*4), 165–232. https://doi:10.1080/15532739.2011.700873

Dalton, E., & Schneider, K. (2014). When to suspect hereditary cancer syndromes. Cancer Consult: Expertise for Clinical Practice, 793–799. https://doi:10.1002/9781118589199.ch123

DeMarco, T. A., Peshkin, B. N., Mars, B. D., & Tercyak, K. P. (2004). Patient satisfaction with cancer genetic counseling: A psychometric analysis of the genetic counseling satisfaction scale. *Journal of Genetic Counseling, 13*(4), 293-304. https://doi:10.1023/b:jogc.0000035523.96133.bc

Edmiston, E. K., Donald, C. A., Sattler, A. R., Peebles, J. K., Ehrenfeld, J. M., & Eckstrand, K. L. (2016). Opportunities and gaps in primary care preventative health services for transgender patients: A systemic review. *Transgender Health, 1*(1), 216-230. https://doi:10.1089/trgh.2016.0019

Erblich, J., Brown, K., Kim, Y., Valdimarsdottir, H. B., Livingston, B. E., Bovbjergm D. H. (2005). Development and validation of a breast cancer genetic counseling knowledge questionnaire. *Patient Education and Counseling, 56,* 182-191. https://doi:10.1016/j.pec.2004.02.007

Frangella, J., Otero, C., & Luna, D. (2018). Strategies for effectively documenting sexual orientation and gender identity in electronic health record. *Studies in Health Technology and Informatics, 247,* 66–70.

Glessner, H. D., VandenLangenberg, E., Veach, P. M., & LeRoy, B. S. (2012). Are genetic counselors and GLBT patients "on the same page"? An investigation of attitudes, practices, and genetic counseling experiences. *Journal of Genetic Counseling, 21*(2), 326-336. https://doi:10.1007/s10897-011-9403-8

Griepsma, M., de Roy van Zuidewijn, D. B., Grond, A. J., Siesling, S., Groen, H., de Bock, G.H. (2014) Residual breast tissue after mastectomy: How often and where is it located? *Annals of Surgical Oncology*. *21*(4):1260-1266. https://doi:10.1245/s10434-013-3383-x

Guillem, J. G., Wood, W. C., Moley, J. F., Berchuck, A., Karlan, B. Y., Mutch, D. G., Gagel, R. F., Weitzel, J., Morrow, M., Weber, B. L., Giardiello, F., Rodriguez-Bigas, M. A., Church, J., Gruber, S., Offit, K., ASCO, & SSO (2006). ASCO/SSO review of current role of risk-reducing surgery in common hereditary

cancer syndromes. *Journal of Clinical Oncology*. *24*(28), 4642–4660. https://doi:10.1200/JCO.2005.04.5260

Harb, C. Y. W., Pass, L. E., De Soriano, I. C., Zwick, A., & Gilbert, P. A. (2019). Motivators and barriers to accessing sexual health care services for transgender/genderqueer individuals assigned female sex at birth. *Transgender Health*, *4*(1), 58-67. https://doi:10.1089/trgh.2018.0022

Hardacker, C.T., Rubinstein, B., Hotton, A., & Houlberg, M. (2014). Adding silver to the rainbow: The development of the Nurses' Health Education About LGBT Elders (HEALE) cultural competency curriculum. *Journal of Nursing Management, 22*, 257–266. https://doi:10.1111/jonm.12125

Haviland, K.S., Swette, S., Kelechi, T., Mueller, M. (2020) Barriers and facilitators to cancer screening among LGBTQ individuals with cancer. *Oncology Nursing Forum, 47*(1):44-55. https://doi:10.1188/20.ONF.44-55

Hughes, C., Lerman, C., Schwartz, M., Peshkin, B. N., Wenzel, L., Narod, S., Corio, C., Tercyak, K. P, Hannah, D., Isaacs, C., & Main, D. (2002). All in the family: Evaluation of the process and content of sisters' communication about BRCA1 and BRCA2 genetic test results. *American Journal of Medical Genetics, 107*(2), 143–150. https://doi:10.1002/ajmg.10110

Johnson, M. J., Mueller, M., Eliason, M. J., Stuart, G., & Nemeth, L. S. (2016). Quantitative and mixed analyses to identify factors that affect cervical cancer screening uptake among lesbian and bisexual women and transgender men. *Journal of Clinical Nursing*, *25*(23-24), 3628-3642. https://doi:10.1111/jocn.13414

Kamen, C. S., Alpert, A., Margolies, L., Griggs, J. J., Darbes, L., Smith-Stoner, M., Griggs, J. J., Darbes, L., Smith-Stoner, M., Lytyle, M., Poteat, T., Scout, N. F. N., & Norton, S. A. (2019). "Treat us with dignity": A qualitative study of the experiences and recommendations of lesbian, gay, bisexual, transgender, and queer (LGBTQ) patients with cancer. *Support Care Cancer, 27*(7), 2525-2532. https://doi:10.1007/s00520-018-4535-0

Mesters, I., van den Borne, H., McCormick, L., Pruyn, J., de Boer, M., & Imbos, T. (1997). Openness to discuss cancer in the nuclear family. *Psychosomatic Medicine, 59*(3), 269–279. https://doi:10.1097/00006842-199705000-00010

Narayan, A., Lebron-Zapata, L., & Morris, E. (2017). Breast cancer screening in transgender patients: findings from the 2014 BRFSS survey. *Breast Cancer Research and Treatment, 166*(3), 875-879. https://doi:10.1007/s10549-017-4461-8

NCCN (2020). National Comprehensive Cancer Network. NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines). Genetic/familial high-risk assessment: breast, ovarian, and pancreatic cancer. Version 1.2021. Available at: https://www.nccn.org/professionals/physician_gls/pdf/genetics_bop.pdf. Accessed October 31, 2020.

O'Connor, A. M. (1995) Validation of a decisional conflict scale. *Medical Decision Making*, *15*(1):25-30. https://doi:10.1177/0272989X9501500105

Ondrusek, N., Warner, E. & Goel, V. Development of a knowledge scale about breast cancer and heredity (BCHK). *Breast Cancer Research and Treatment, 53*, 69–75 (1999). https://doi:10.1023/A:1006114710328

Patterson, J. G., Jabson Tree, J. M., & Kamen, C. (2019). Cultural competency and microaggressions in the provision of care to LGBT patients in rural and Appalachian Tennessee. *Patient Education and Counseling, 102*(11), 2081-2090. https://doi:10.1016/j.pec.2019.06.003

Rafferty, J. (2018). Ensuring comprehensive care and support for transgender and gender-diverse children and adolescents. *Pediatrics, 142*(4), 1-15. https://doi:10.1542/peds.2018-2162

Read, C. Y., Perry, D. J. and Duffy, M. E. (2005), design and psychometric evaluation of the psychological adaptation to genetic information scale. *Journal of Nursing Scholarship, 37*: 203-208. https://doi:10.1111/j.1547-5069.2005.00036.x

Sacca, R. E., Koeller, D. R., Rana, H. Q., Garber, J. E., & Morganstern, D. E. (2019). Trans-counseling: A case series of transgender individuals at high risk for BRCA1 pathogenic variants. *Journal of Genetic Counseling, 28*(3), 708-716. https://doi:10.1002/jgc4.1046

Sheehan, E., Bennett, R.L., Harris, M., Chan-Smutko, G. (2020) Assessing transgender and gender non-conforming pedigree nomenclature in current genetic counselors' practice: The case for geometric inclusivity. *Journal of Genetic Counseling.* https://doi:10.1002/jgc4.1256

Shiloh, S., Avdor, O., & Goodman, R. M. (1990). Satisfaction with genetic counseling: Dimensions and measurement. *American Journal of Medical Genetics, 37*(4), 522–529. https://doi:10.1002/ajmg.1320370419

Stalmeier, P. F., Roosmalen, M. S., Verhoef, L. C., Hoekstra-Weebers, J. E., Oosterwijk, J. C., Moog, U., Hoogerbrugge, N., & van Daal, W. A. (2005). The decision

evaluation scales. *Patient Education and Counseling, 5*7(3), 286–293. https://doi:10.1016/j.pec.2004.07.010

Sutherland, N., Espinel, W., Grotzke, M., & Colonna, S. (2020). Unanswered questions: Hereditary breast and gynecological cancer risk assessment in transgender adolescents and young adults. *Journal of Genetic Counseling,* 29(4), 625–633. https://doi:10.1002/jgc4.1278

Tabaac, A.R., Sutter, M.E., Wall, C.S.J., & Baker, K.E. (2018). Gender identity disparities in cancer screening behaviors. *American Journal of Preventive Med, 54*, 385–393. https://doi:10.1016/j.amepre.2017.11.009

Tuite, A., Piazza, M. D., Brandi, K., & Pletcher, B. A. (2020). Beyond circles and squares: A commentary on updating pedigree nomenclature to better represent patient diversity. *Journal of Genetic Counseling*, 29(3), 435–439. https://doi:10.1002/jgc4.1234

VandenBos, G. R., & American Psychological Association. (2007). *APA dictionary of psychology*. Washington, DC: American Psychological Association. https://dictionary.apa.org/

VandenLangenberg, E., Veach, P. M., LeRoy, B. S., & Glessner, H. D. (2012). Gay, lesbian, and bisexual patients' recommendations for genetic counselors: A qualitative investigation. *Journal of Genetic Counseling, 21*(5), 741-747. https://doi:10.1007/s10897-012-9499-5

von Vaupel-Klein, A. M., & Walsh, R. J. (2020). Considerations in genetic counseling of transgender patients: Cultural competencies and altered disease risk profiles. *Journal of Genetic Counseling*. https://doi:10.1002/jgc4.1372

Warren, N.S., & Wilson, P.L. (2013). *A 10-point approach to cultural competence in genetic counseling*. Genetic Counseling Cultural Competence Toolkit. http://www.geneticcounselingtoolkit.org/COUNSELING-PGC%202013.pdf

Zayhowski, K., Park, J., Boehmer, U., Gabriel, C., Berro, T., & Campion, M. (2019). Cancer genetic counselors' experiences with transgender patients: A qualitative study. *Journal of Genetic Counseling, 28*(3), 641-653. https://doi:10.1002/jgc4.1092

APPENDIX A

PARTICIPANT RECRUITMENT SOCIAL MEDIA TEXT

The following text was posted to various social media platforms with a direct link to the survey.

Jacqueline Baquet, a genetic counseling student at the University of South Carolina, is working on a thesis project to assess the needs of transgender individuals in a cancer genetic counseling setting. This is an online survey for individuals whose gender identity does not align with their gender assigned at birth.

Please consider participating if any of the following applies to you:

- Have or have had cancer yourself
- Have relatives that have or have had cancer
- Are interested in learning about your cancer risks
- Are interested in learning about genetic testing for hereditary cancer
- Are interested in contributing to research that will benefit this healthcare field

Please share this post with anyone who may be eligible to participate in this survey.

Upon completion of the survey, you will be offered the option to enter a raffle for one of five $25 Visa gift cards as compensation for your time.

APPENDIX B

QUESTIONNAIRE

The following list of questions was used as an outline for the questionnaire after inclusion criteria was met.

Where appropriate, the interviewees were asked to expand upon their answers.

1. Please select the gender(s) that best describe you:

2. Have you ever been diagnosed with cancer?

i. What type of cancer(s) have you been diagnosed with?

ii. How old were you when you were diagnosed with cancer?

3. How much do you feel you know about your family medical history?

4. Please select the number of relatives that have been diagnosed with the following cancers:

i. Breast cancer

ii. Ovarian cancer

iii. Gastric/stomach cancer

iv. Colon/rectal cancer

v. Pancreatic cancer

vi. Melanoma

vii. Prostate cancer

viii. Endometrial/uterine cancer

ix. Other cancer

x. Unsure what type of cancer

4. Please select a response to the following considerations regarding a cancer genetic counseling appointment:

i. If I was at an increased risk to develop cancer due to a genetic cause, I would feel comfortable sharing this with my relatives without assistance from a genetic counselor.

ii. If I was at an increased risk to develop cancer due to a genetic cause, I would feel comfortable sharing this with my relatives with assistance from a genetic counselor like a family letter, phone call, or follow-up appointment.

iii. It is important to me that medical intake forms ask about gender identity in an inclusive way.

iv. It is important to me that medical professionals exchange pronouns with me.

v. It is important to me that medical professionals use inclusive language during an appointment.

vi. It is important to me that I am represented in my medical records as my gender, rather than my gender assigned at birth.

vii. It is important to me that medical offices display signs or other visual representations of LGBTQ+ inclusivity in their waiting room.

5. Please select a response to the following considerations regarding a cancer genetic counseling appointment:

i. I would prefer being asked directly about my pronouns over voluntarily sharing them.

ii. I would feel comfortable sharing my pronouns on an intake form before my genetic counseling appointment.

iii. I would feel comfortable sharing my pronouns with my genetic counselor as soon as I am called from the waiting room.

iv. I would feel comfortable sharing my pronouns with my genetic counselor when I am inside their office.

v. I would feel comfortable sharing my pronouns after the genetic counselor shared theirs.

vi. I would feel comfortable disclosing any gender-affirming hormones and surgeries I've had on an intake form before my genetic counseling appointment.

vii. I would feel comfortable disclosing any gender-affirming hormones and surgeries I've had during my genetic counseling appointment.

viii. I would feel comfortable being represented on the official pedigree as my gender identity with AFAB/AMAB written underneath.

ix. I would feel comfortable being represented on the official pedigree as some combination of my assigned sex and gender identity.

x. I would feel comfortable being represented on the official pedigree as my sex assigned at birth.

6. Please select a response to the following considerations regarding a cancer genetic counseling appointment:

i. I would value learning about my personal cancer risk.

ii. I would value learning about the cancer risk for my children.

iii. I would value learning about the cancer risk for my relatives.

iv. I would value having someone to help me understand how my personal and family history impacts my cancer risk.

v. I would value discussing my thoughts and feelings about my cancer risk.

vi. I would value discussing my thoughts and feelings about my genetic test results.

vii. I would value discussing my thoughts and feelings about cancer screening procedures.

viii. I would value discussing how my cancer risk could impact my family planning decisions.

ix. I would value discussing hormone therapies and gender-affirming surgeries as they relate to my cancer risk.

x. I would value having someone to help me understand my options for cancer detection.

xi. I would value having someone to help me understand my options for cancer prevention and risk-reduction.

xii. I would value having someone to help me decide if genetic testing is right for me.

xiii. I would value discussing costs relating to genetic testing and follow-up screening.

xiv. I would value having someone to help me understand the privacy and confidentiality policies surrounding genetic test results.

7. Have you ever taken, or considered taking, gender-affirming hormones?

8. Have you ever had, or considered having, surgery on your breast tissue?

9. Were you born with ovaries?

    i. Have you ever had, or considered having, surgery to remove your ovaries?

10. Were you born with a uterus?

    i. Have you ever had, or considered having, surgery to remove your uterus?

11. Were you born with a prostate?

    i. Have you ever had, or considered having, surgery to remove your prostate?

12. What is your age?

13. Please select the race(s) that best describe you:

14. Please select the ethnicity that best describes you:

15. Please select the highest level of education you have completed:

16. Please select your employment status:

17. Please select the annual income that best describes your household:

18. Please select your current health insurance:

19. Do you currently receive care from a health center that specializes in health care for transgender individuals?

APPENDIX C

PHONE INTERVIEW

The following list of questions was used as an outline for the phone interview questions.

1. What are your pronouns?

2. How do you feel about discussing cancer risks with your family?

3. What about with your doctors?

4. Which of your doctors have asked you about your cancer family history?

i. What did they say?

ii. Is this something you wish would have been addressed?

5. Has any doctor ever talked to you about cancer as it relates to any hormones you have taken or surgeries you have had?

i. What did they say?

ii. Is this something you wish would have been addressed?

6. Can you tell me about what you have been told regarding cancer screening?

7. How do you feel about cancer screenings?

    i. Have you had any cancer screenings yourself? What type? Why not?

8. Do you think you might feel differently if you knew you were at an increased risk for developing cancer?

9. What makes you most comfortable when visiting a new healthcare office?

10. What do you see as the largest benefit of pursuing cancer genetic counseling? What would be your largest reservation about pursuing cancer genetic counseling?