

**Genetic Counseling for Trans* Populations: Development of Educational Material for
Genetic Counseling Programs**

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Abstract

Transgender, or trans* is an umbrella term that refers to people whose gender identity differs from the sex they were assigned at birth. As gender minorities, the trans* community experiences disproportionate health disparities and discrimination in the healthcare setting. Genetic counselors may become part of a trans* individuals' medical care team when indicated by a risk for genetic conditions. The role of a genetic counselor is to provide genetic information and culturally competent psychosocial support. A recent survey of genetic counselors found that less than half felt confident in their ability to counsel a trans* client, and multiple studies have established genetic counselors' desire for more education on counseling trans* individuals. This study aims to increase genetic counselors' knowledge about counseling trans* clients through the creation of educational resource materials for genetic counseling training programs accredited by the Accreditation Council for Genetic Counseling (ACGC). A Qualtrics survey was distributed to ACGC accredited program leadership to assess their current approach to trans* health education within their curricula and their receptiveness to additional resources to increase students' exposure and comfort level counseling trans* patients in the future. Survey data revealed that 90% of responding programs provide education on genetic counseling for trans* individuals, and 85% felt that access to educational resource materials would be helpful. The results of the survey were used to direct what kind of educational content was created as part of this project. Based on survey results, four resources were created: 1) A series of case examples with roleplay opportunities, 2)

An annotated list of resources for trans* patients including allied healthcare providers and legal resources, 3) An annotated list of resources for providers working with trans* clients, 4) An annotated list of peer-reviewed articles on genetic counseling for trans* populations. An educational resource guide was made to guide users of these materials as well. This study is relevant to public health by increasing educational materials available to genetic counseling programs and their students, with the intention of enhancing genetic counselors' competency in working with trans* clients.

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Preface

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1.0 Introduction

1.1 Background and Specific Aims

Trans* individuals are a growing demographic in the United States, some of whom have unique healthcare needs.¹ A transgender person is someone whose gender identify is incongruent with, or socially perceived to be different, from their sex assigned at birth (SAAB). The trans* umbrella term includes individuals who identify with the binary sex opposite that that they were assigned at birth as well as those who identify outside the binaries of man and woman. In 2016, it was estimated that 1.4 million adults in the United States identified as transgender, which does not account for transgender youth, and may underestimate those with non-binary gender identities.² Studies including transgender youth have found that the proportion of individuals who identify as trans* are higher in youth populations than adult populations, possibly as a result of increasing awareness, acceptance, and self-reporting.¹ Many medical professionals such as genetic counselors already encounter trans* individuals in clinical settings. As the trans* population in the United States grows, it becomes more likely that genetic counselors will encounter trans* clients during their careers.

Multiple organizations such as the American Public Health Association and the 2020 Healthy People campaign have recognized the need for improvement in the health disparities trans* people in America face.^{3,4} Mental health concerns such as depression and anxiety appear to be elevated in trans* populations.⁵ A national survey of trans* individuals revealed that 41% had tried to commit suicide, which is four times the national average.⁵ Trans* individuals are more likely than cisgender individuals to experience physical assault, sexual assault, intimate partner

violence, and are much more likely to delay important medical treatment.⁶ In addition, trans* people are more likely to be uninsured, and experience unemployment, homelessness, legal discrimination in adoption and marriage, and loss of support from parents or family members.⁵

Healthy People 2020, a national initiative focused on using scientific data to set goals for improving the health of Americans, cited a lack of knowledgeable providers for lesbian, gay, bisexual, and transgender (LGBT) individuals as one of the top barriers negatively affecting the health outcomes of trans* people.⁴ Nearly a quarter of trans* individuals report having been refused care, and over a quarter had experienced harassment in a healthcare setting.⁵ A trans* person is more likely to experience discrimination from a healthcare provider when their trans* identity is disclosed, despite studies suggesting healthcare providers need this information to provide appropriate care.⁵ This suggests that disclosing gender identity will improve a person's care only if the provider receiving the disclosure is well educated on trans* health and respectful towards trans* identities.⁷ Overall, of those who had seen a healthcare provider in the last year, one-third of trans* people reported at least one experience of harassment or discrimination with their healthcare provider which was related to their trans* status.⁶

Clinical medical professionals are tasked with providing competent and compassionate care to all of their patients of all backgrounds. Genetic counselors facilitate “the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.”⁸ Genetic counselors are expected to understand genetic and psychosocial considerations unique to each patient, provide culturally competent counseling, and establish rapport with clients using counseling strategies. Genetic counseling has the potential to benefit trans* individuals seeking guidance on their risk for genetic conditions, reproductive risks and options, and medical decision making. While there is a benefit for genetic counselors to provide

informed care, gaps in care persist, especially among racial, sexual, and gender minority populations.⁹ Genetic counselors are professionals who often provide resources to clients facing many of the psychological, social, and financial disparities many trans* individuals do at disproportionate rates.¹⁰

This study elicited current information on the structure of curricular content related to genetic counseling for trans* individuals in ACGC-accredited genetic counseling programs, as well as any barriers to providing this education. This study also assessed gaps in education and any educational resources that would be helpful for programs to have access to on the topic. The results of this study were used to guide the development of educational resource materials designed to increase the knowledge base of genetic counseling trainees. Ultimately, increasing genetic counselors' knowledge regarding genetic counseling issues may increase a genetic counselors' comfort and improve the quality of care for trans* clients.

1.1.1 Specific Aim 1

Design and distribute a Qualtrics survey to ACGC-accredited genetic counseling programs to assess current curricula in genetic counseling for transgender and gender nonconforming (GNC) individuals, as well as programs' perception of educational resource material on this topic.

1.1.2 Specific Aim 2

Use the survey data, primary literature involving perspectives from transgender and GNC individuals, and best practices guidelines from other healthcare professions and LGBT health

organizations to develop and distribute educational resource materials for genetic counseling programs.

2.0 Literature Review

2.1 Defining the population

The growing list of terms referring to gender and sex can be daunting to providers who should aim to use the most accurate terms to discuss and address their patients. Because of the historical marginalization of sex and gender minority patients, it remains paramount for the patients themselves to self-disclose and be empowered to use terms that best describe their experience. While this self-expression is needed to work towards societal acceptance and gender literacy, a common language among healthcare providers is needed for successful communication, provision of healthcare, and information gathering.¹¹ In an effort to operationalize common terms used in the discussion of issues pertaining to sex and gender identity, a list of terms is provided in Table A.

Table A: Relevant Terms

Term	Definition
Sex assigned at birth (SAAB) (n.)	A designation of male or female (as well as intersex in some states) assigned to babies at birth usually based on external genitalia
Gender identity (n.)	An internal and personal understanding of one's own gender which may or may not correspond with biological sex
Gender expression (n.)	A person's presentation of gender visible to the outside world through a person's preferred name, pronouns, clothing, haircut, behavior, and voice, oftentimes designated as masculine or feminine
Transgender (adj.)	A descriptor for a person whose gender identity is incongruent with or different from the sex they were assigned at birth
Cisgender (adj.)	A descriptor for a person whose gender identity is congruent with or the same as their sex assigned at birth
Disorders of sex development (n.)	A group of medical conditions that result in a mismatch between a person's chromosomes and internal/external genitalia as well as secondary sex characteristics
Gender nonconforming (GNC) (adj.)	A descriptor for individuals whose behavior, physical traits, cultural norms, and/or presentation of gender is socially perceived to be incongruent with the sex they were assigned at birth
Nonbinary (adj.)	An umbrella term for individuals whose gender identity is not described by the binary identities of man and woman
AMAB (adj.)	An acronym for assigned male at birth—refers to a person whose assigned biological sex is male

AFAB (adj.)	An acronym for assigned female at birth—refers to a person whose assigned biological sex is female
MtF (adj.)	An acronym for male to female—generally refers to a person who was assigned male at birth but identifies as female in gender identity and/or expression
FtM (adj.)	An acronym for female to male—generally refers to a person who was assigned female at birth, but identifies as male in gender identity and/or expression
Transition (v.) (n.)	A personal process of making changes to one's appearance through measures such as changes in personal style, hormone therapy and gender affirming surgery to better align one's physical self with gender identity
Gender dysphoria (n.)	A psychologically painful phenomenon produced by the discord between one's gender identity and sex assigned at birth
Trans* (adj.)	A term that has emerged to describe all individuals who are not cisgender, including identities outside of the gender binary
Gender neutral (adj.)	A descriptor for a word or expression that can be applicable to all genders—usually implies that no one gender is specified

2.1.1 Sex, gender identity, and gender expression

In general, a person is assigned a sex at birth—either male, female, or in some cases, intersex. In general babies with vaginas are assigned female, and babies with penises are assigned male. In rare circumstances the genetic sex of baby may be known and may inform sex assignment.

While sex may have a biological basis with its own spectrum of variability, gender identities are socially constructed self-concepts that are often separate from the biological sex assigned at birth (SAAB).¹² Gender identity refers to a person's internal sense of gender. This relies on whether a person sees themselves as a man, woman, both, or neither, and need not be related to a person's sex assigned at birth.¹¹ Often, but not always, a person's gender identity influences which pronouns a person feels describes them best. For instance, someone who identifies as a man may use "he," "him," "his," while someone who identifies as a woman may use "she," "her," and "hers." Among other possible preferences, there is the gender-neutral option of using "they," "them," and "theirs." The sex a person is assigned at birth operates as a medical and legal identity, while gender identity operates as a social identity and affects how a person thinks about themselves and experiences the world.¹³

Gender identity or sex cannot be assumed based on first impressions. How individuals present themselves and their gender to the world is considered their gender expression. This may or may not coincide with the gender identity that others may assume for them.¹⁴ Generally, gender expression is viewed on the spectrum of masculine to feminine. One way to conceptualize the difference between gender expression and gender identity is that gender expression is an external expression, while gender identity is an internal understanding of one's gender.¹⁵ Western ideas about sex, gender identity, and gender expression tend to operate on a binary system, where a person can be either male or female, a man or a woman, masculine or feminine. However, there are many gender identities that fall outside of this binary conceptualization, such as those who identify as nonbinary, genderqueer, or agender.¹⁴ Individuals with these identities may see themselves as neither a man nor a woman, or as a gender entirely different from either of those

categories. Individuals within this population, along with individuals of all other gender identities, may express their gender in a gender neutral or gender nonconforming (GNC) fashion.

2.1.2 Transgender and cisgender

A person is cisgender when their gender identity aligns with or is the same as their sex assigned at birth (SAAB). A person is considered to be transgender when their gender identity is incongruent with or socially perceived to be different from their SAAB. In 2016, an estimate was made that 1.4 million adults in the United States identified as transgender, which does not account for transgender youth, and may underestimate those who identify with non-binary gender identities.² Some but not all transgender/GNC individuals may experience gender dysphoria, a psychologically painful mental condition produced by the discord between one's sense of gender identity and their SAAB.¹⁶ Gender dysphoria is not a mental illness in of itself, but can be a source of ongoing mental health issues and distress. Some but not all transgender/GNC individuals seek out gender affirming treatments such as hormone therapy and gender affirming surgeries.¹⁷ Presence of gender dysphoria and desire for surgery do not always correlate with one another.¹⁶

A transgender person generally will have been assigned male at birth (AMAB) or assigned female at birth (AFAB), but not identify with the sex they were assigned at birth as being representative of their gender identity. Though many transgender identities exist outside of binary understandings of gender, generally someone who is AMAB and identifies and lives as a woman would be a transgender woman, often denoted as being male to female (MtF). Likewise, someone who is AFAB and identifies as a male would be a transgender man, or female to male (FtM). The terms “transgender man” and “transgender male,” as well as “transgender woman” and “transgender female” are often used interchangeably, however this document will use “transgender

man” and “transgender woman” to describe gender identities in order to avoid confusion of these terms with sex categories.

2.1.3 Trans* identities and population of interest

As mentioned previously, there are many terms for transgender and gender nonconforming identities. Trans* is a term that has emerged in recent years to describe all individuals who do not identify as a cisgender. Though transgender and trans have also been used as blanket terms for this population in the past, supporters of the use of trans* argue that the term provides more inclusivity of identities that are outside of a binary understanding of gender. The asterisk symbolizes the diversity of gender identities within the trans* umbrella. This creates a term for a community that is less medicalized and more inclusive of many individuals’ understanding of their own gender.^{18,19} For the purposes of this study, trans* will be used to describe all non-cisgender individuals, as the information herein extends to all non-cisgender individuals and transgender people outside of binary transgender identities.

Importantly, this study does not specifically address education for genetic counselors in counseling individuals with disorders of sex development or differentiation (DSDs), which are medical conditions that affect individuals’ sexual development.²⁰ Though some individuals living with a DSD may also identify as trans*, many do not. Even those who may have been designated intersex at birth may not identify with “intersex” as encapsulating their gender identity. There are many unique considerations for this population for genetic counselors to be aware of that are outside the scope of this study.

2.2 Defining the problem

Lesbian, Gay, Bisexual, and Transgender (LGBT) individuals often experience disparities and discrimination in healthcare due to their sexual and gender minority status. Trans* individuals experience different barriers from others in the LGBT community, in part due to more interactions with and dependence upon the healthcare system for transition-related purposes,²¹ and because transphobia and general lack of awareness for care of transgender individuals is pervasive.⁵ To fully define the barriers facing this population for competent care, it's important to consider multiple perspectives; from a trans* person's ability to access care and their interactions with providers to the systemic barriers providers face in offering more competent care to this population.

2.2.1 Health disparities

In 2000, the American Public Health Association released an official policy statement acknowledging the healthcare disparities in transgender populations and stating the organization's desire to improve treatment.³ Twenty years later, the Healthy People campaign still identifies LGBT health, safety, and well-being as an area of public health in need of serious improvement.⁴

LGBT populations experience higher rates of substance use, tobacco use, and obesity than the population average.⁴ Mental health concerns such as depression and anxiety appear to be elevated specifically in the trans* population,⁵ as 41% of trans* respondents to a national survey reported attempting suicide—four times the national average.⁵ Additionally, trans* individuals are more likely to experience physical assault, sexual assault, intimate partner violence, and are much more likely to delay important medical treatment than cisgender individuals out of fear of

discrimination.⁶ Furthermore, trans* people are more likely to be uninsured, experience unemployment, homelessness, legal discrimination in adoption and marriage, and loss of support from parents or family members, all of which are considered to be social determinants of health.⁵

2.2.2 Competency of care from providers

Despite public health efforts to improve trans* healthcare, problems at the provider level still need to be addressed. Healthy People 2020 cited a lack of knowledgeable providers for LGBT individuals as one of the top barriers negatively affecting the health of trans* people.⁴ In one study, access to a provider who was knowledgeable on trans* health as well as access to a provider who was trans*-friendly were reported as the top two barriers to healthcare that trans* people faced, even more common than significant barriers associated with healthcare cost and distance from clinics.²² Many trans* individuals have explained that the barrier of having to travel to faraway clinics for care often stems from providers who are incompetent in trans* care or outright refuse to see trans* patients more locally.²³ Nearly a quarter of trans* individuals report having been refused care, and over a quarter had experienced harassment in a healthcare setting.⁵ Though studies suggest that knowledge of both sex and gender identity is vital for a provider to have in order to deliver appropriate care,¹¹ a trans* person is more likely to experience discrimination from a healthcare provider when their trans* status is known to them.⁵ This suggests that disclosing one's gender identity will improve a person's care only if the provider receiving the disclosure is educated on providing care for trans* clients.⁷ Of those who had seen a healthcare provider in the last year, one-third of trans* people reported at least one experience of harassment or discrimination with their healthcare provider which stemmed directly from their trans* status.⁶

Independent of blatant discrimination and harassment, many providers are ill-equipped to provide competent care to trans* patients. In a national survey of trans* individuals in America, half reported that they had to teach their provider about their trans* identity and associated healthcare needs.⁵ Additionally, a majority of trans* people report at least one instance of a provider using gendered language or making assumptions which made them feel uncomfortable.²⁴ The Joint Commission asserts that a provider's failure to obtain information on a patient's sexual orientation and gender identity should be considered akin to a failure to diagnose,²⁵ yet almost one-third of trans* individuals report having never disclosed their trans* status to any of their providers.⁶ Many studies have found that professionals are aware of their shortcomings in care of this population.⁶ A recent study of gynecologists found that up to two thirds felt uncomfortable caring for transgender men and women.²⁶ Based on reports from trans* individuals, this general discomfort seems to extend to clinical encounters and often preclude negative outcomes.⁶ Even the well-intentioned provider can hurt their relationship with their patient by not using the right language and terms

2.2.3 Systemic barriers in healthcare

Further complications arise for both patients and providers trying to navigate a healthcare system not built for trans* people. The medical field is entrenched in a binary and static approach to gender.¹³ Many trans* individuals have problems with the healthcare system not just because of encounters with undereducated and biased providers, but because the medical system separates trans* people from the general population as special cases or exceptions to the rule.²⁷ Because of this narrow view, even well-informed providers may not be able to implement appropriate services when working in a clinical system that does not have trans* inclusive intake forms, online record

systems, or anti-discrimination policies in place.¹³ Many healthcare sites still employ restrictive documentation policies when it comes to a person's SAAB, and little trans* health education is done in professional programs and ongoing education efforts, which makes understanding trans* healthcare needs the sole responsibility of the individual provider.¹³ While many providers do advocate for systemic change within their medical setting, these changes can take time. Additionally, many providers cite that identifying competent referral networks for trans* patients is difficult to impossible, leading to anxiety about the competency of the referral.¹³ Official guidelines regarding treatment for trans* individuals, though currently available for many specialties, often fail to address or inform many decisions providers must make and are based on little data, causing many providers to feel unsure and ill-prepared even after working to educate themselves.¹³ These systemic barriers enhance negative trans* healthcare experiences and make provision of trans* sensitive healthcare more difficult for providers to employ.

2.3 Identifying what is known: trans* healthcare

Research to learn more about trans* healthcare needs and efforts to educate healthcare professionals exist, both within the medical community and from trans* advocates. Though gaps in provider knowledge and available data exist, resources from trans* advocacy organizations and other clinical specialties have the potential to provide valuable information to healthcare providers. Having a good understanding of what is already known in the field of trans* health is key to identifying what information might be important to include when educating future providers.

2.3.1 Clinical guidelines and educational resources for healthcare professionals

Increasingly, educational resource material has become available to practicing clinicians and clinical training programs for the appropriate treatment of trans* patients. Many professional institutions and advocacy organizations have released trainings, guidelines, and best practices for treating trans* patients and for educating young professionals about trans* healthcare.^{16,25,28-35} While the National Society of Genetic Counselors has provided webinars on LGBT health issues in the past, no official guidelines for care of trans* patients have been published for genetic counseling. Though the majority of existing training tools and guidelines were created with other specialties in mind, many contain recommendations and information that can be applied to genetic counseling.

2.3.1.1 Clinical guidelines and educational resources from professional health organizations

In 2010, the American Counseling Association released their Competencies for Counseling with Transgender Clients,²⁸ and in 2015, the American Psychological Association released Guidelines for Psychological Practice with Transgender and Gender Nonconforming People.¹⁶ These documents outline suggested competencies for all counselors to master in order to appropriately provide care to trans* patients. Though some of the suggested competencies lend themselves better to psychological counseling over a longer period of time, many of the recommendations can be applied to genetic counselors in their psychosocial assessment and care of a patient, such as the recommendation that counselors be adequately prepared to discuss differing family dynamics and the possibility of estrangement. Other psychologists have attempted to explain and recommend trans* friendly counseling strategies through the lens of the Minority Stress Model, a widely adopted way of conceptualizing sexual and gender minority oppression.³⁶

The American Academy of Family Physicians (AAFP) has a published recommended curriculum for family medicine residents for LGBT health issues and provision of care which contains fairly comprehensive recommendations on education for trans* health issues.²⁹ A portion of the recommendations for education is rooted in allowing students to acknowledge their own preconceptions about gender and sex, and gives recommendations on how these conversations should be mediated by the instructor. There are some key recommendations regarding care of the trans* patient that are applicable to all healthcare providers. Additionally, the Association of American Medical Colleges (AAMC) website has a wealth of educational resources available to educators that are designed to assist them in having conversations with students about trans* issues, such as one that advises educators on how to teach students the concept of gender identity versus biological sex.³¹ Another AAMC resource provides information to practitioners on how to accurately and competently talk to patients and elicit information on sexual orientation/gender identity.³⁰ Additionally, groups of trans* and trans* friendly providers have created resources for their own colleagues online, such as OutCare, a database that assists trans* individuals with finding competent providers, and helps providers to more easily find and refer trans* patients to specialists who have experience in providing competent care to trans* people.³⁷

The Joint Commission, a professional organization which exists to create standards and accreditations in healthcare in order to inspire all providers to continually improve their knowledge and quality of care, published their official standards of care for LGBT patients in 2011, touching on the need for cultural competence, communication, and patient/family-centered care.²⁵ Though much of the document talks about hospital policies, a portion of the document is dedicated to recommendations on clinical care for LGBT patients, including the use of gender-neutral language, consistent and competent documentation of SO/GI (sexual orientation/gender identity), and the

need to become familiar with community resources available to these populations.²⁵ AHIMA, another organization which sets healthcare standards specifically when it comes to managing healthcare data, published a set of recommendation and standards regarding health information management for individuals who are LGBT.³² This document emphasizes the responsibility of healthcare providers to competently obtain SO/GI information from patients, as well as ensure the patient is aware of the privacy of this information. Importantly, the resource also discusses the need for more education on these topics in the healthcare setting, and addresses the importance of being honest with LGBT patients about possible gaps in competent record-keeping and current technology.³² Currently established clinical guidelines within other medical specialties contain important considerations that can be applied to a genetic counseling session, and multiple studies on more specific trans* healthcare issues that have been used by professional healthcare organizations that directly relate to some genetic counseling specialties, such as one on fertility options for trans* individuals.³⁸

2.3.1.2 Clinical guidelines and educational resources from trans* and patient advocacy organizations

Trans* advocacy organizations play an important role in informing the public of trans* issues, as well as their history. Many groups have published guidelines or recommendations for healthcare providers on how to best serve the population. The Center of Excellence for Transgender Health at the University of California San Francisco has created coursework for medical professionals including educational videos, perspectives from transgender individuals, and the opportunity for self-assessment of learned skills in providing care for transgender individuals in the form of case studies.³⁴ These materials have been endorsed by the CDC, who have encouraged the adoption of these materials into healthcare curriculum. The National Center

for Transgender Equality has also created resources not only for providers, but for trans* individuals navigating the medical system as well, such as their “Know Your Rights” piece which outlines the legal protections for trans* individuals in healthcare, also widely distributed to healthcare providers who should be familiar with these protections.³³ The World Professional Association for Transgender Health (WPATH) has released their own standards of care for the health of trans* individuals, in which they provide clear clinical guidelines formulated by trans* advocates for the healthcare of trans* individuals.³⁵ Though these and other standards and recommendations produced by advocacy groups largely coincide with and inform the standards published by professional health organizations, recommendations coming straight from trans* advocacy groups may be more tailored to trans* healthcare needs than standards created for the LGBT community at large.

2.3.2 Lessons from research within trans* populations

Though many of the guidelines from professional organizations have been informed by trans* patients and healthcare workers, it’s important to be aware of the perspectives from research which examines the perspectives of trans* individuals. Research within trans* populations has allowed individuals seeking to provide competent care a look into the opinions from those who are experts on the topic—trans* individuals themselves. One such study interviewed a large sample of trans* individuals about their experiences with healthcare providers during the process of medically transitioning.²¹ After compiling perspectives from their sample of trans* individuals, the researchers found that when facing healthcare providers, “encounters were perceived [by trans* people] as good when healthcare professionals showed respect and preserved the transsexual person’s integrity, acted in a professional manner and were responsive and built trust and

confidence."²¹ This same sample of individuals felt that healthcare encounters were negative when the provider had a lack of knowledge, exploited their position of power over the patient, used stereotyping language, used incorrect names or pronouns, and more.²¹ Another similar study used LGBT focus groups with the goal of creating a list of assets of and strategies for creating a culturally competent clinical environment for these groups. The aspects of a culturally competent environment fell into three main categories—structural, related to the physical space of the clinic, interpersonal, which dealt with providing comfort and forming a working relationship with patients, and systemic, which involved the mission statements, intake forms, and policies of the clinic at large.³⁹ Guides to competent care of trans* patients have made recommendations on caring for trans* patients not only while the patient is present in the clinic, but also in personal spaces and, importantly, as an advocate for these individuals to insurance companies and other providers.⁴⁰⁻⁴² Though this is not an exhaustive list of the recommendations from trans* individuals that have come out of research, these provide valuable perspectives from those who understand firsthand the impact a competent (or incompetent) healthcare provider can have.

2.3.3 Persisting gaps in knowledge

Despite the knowledge we have about trans* healthcare, gaps in education and research continue to affect the treatment trans* people receive from healthcare providers. Studies have shown reluctance from LGBT individuals to participate in research due to surveys and paperwork lacking in inclusivity, which may cause these unknowns to persist in clinical research.¹⁵ Despite the publication of clinical guidelines in many specialty areas, a recent study found that up to a third of all medical schools currently have no hours of LGBT health curriculum.⁴³ This, in addition to the fact that many standard educational materials lump healthcare for trans* individuals in with

the education needed to serve LGB individuals without addressing issues specific to gender minorities,²¹ demonstrates that the vast majority of clinical healthcare providers may not have relevant information to provide appropriate trans* healthcare despite its increased availability. Overall, the knowledge we have is incomplete, and what we do have may not be reaching providers in educational settings.

2.4 Genetic counseling for trans* individuals: what we know

Genetic counseling is defined by the National Society of Genetic Counselors (NSGC) as “the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.”⁸ Genetic counselors are trained both in genetics and counseling skills in order to help educate patients and other providers on genetic testing options and genetic diseases while taking care of patients psychosocially. Clinical genetic counseling is a rapidly growing service that is becoming more widely accessible. While many efforts have been made to promote and teach multicultural competency in genetic counseling, like all other forms of healthcare in the United States, inequities in access to genetic counseling and quality of care persist for minority populations.⁹ Both because of the lack of research on trans* individuals in healthcare and because of the relative newness of this emerging field, there is not a wealth of literature which explores genetic counseling specifically for trans* individuals. However, a number of papers published in recent years have begun to provide information on how genetic counselors can best serve this population.

Though research on genetic counseling for trans* individuals is still developing, several studies have provided vital insight into trans* perspectives on the healthcare service and how

genetic counselors can best serve them. One recent study conducted with a large sample of trans* individuals found that if referred for genetic counseling, 68% would be worried that the counselor would discriminate against them for their gender identity.⁴⁴ Though the majority of these participants had not previously seen a genetic counselor, and the rate of fear of discrimination among trans* patients was actually higher for physicians (79%),⁴⁴ it is important to recognize how trans* individuals' lived experiences, both with genetic counselors and other providers, shape the way they interact with the healthcare system. Having a clearer understanding of trans* individuals' perspectives on genetic counseling can allow counselors to prepare for these sessions and develop strategies to build rapport with a patient population whose members have historically faced discrimination in healthcare at an increased rate.

Work has been done within the genetic counseling community to assess counseling biases as well as strategies already used by counselors for counseling trans* patients. The goal of one study in particular was to elicit unique considerations during cancer genetic counseling visits with trans* individuals from the perspective of 21 cancer genetic counselors who had seen trans* patients in clinic.⁴⁵ The study identified unique discussions that may need to occur with trans* patients in cancer genetics such as the role of pathogenic genetic variants in insurance coverage for gender-affirming surgeries, as well as recommendations on appropriate language use and identifying systemic problems, such as non-inclusive electronic medical record systems, which genetic counselors found had been barriers to providing competent care to their trans* patients.⁴⁵ Another larger survey of genetic counselors from all specialties and with varying levels of counseling experience with LGBT populations found similar perspectives and barriers and revealed that nearly three out of four (71.8%) counselors' approach to counseling does not differ between LGBT and heterosexual/cisgender clients.⁴⁶ Many of those counselors who reported that

they would approach counseling the same way for both groups reported that they would do so because they believed that this was a method of promoting equal treatment for all clients.⁵⁰ While this perspective is well-intentioned, this information suggests that some practitioners may not be aware of or taking into account the considerations which counselors who work with trans* individuals feel need to be made specifically for this group. Those counselors who reported approaching cisgender clients differently than trans* clients cited that trans* individuals may have different motivations for genetic counseling and may have differing psychosocial needs.⁴⁶

Another survey of cancer genetic counselors sought to establish the confidence and competence of cancer genetic counselors to counsel trans* patients. The survey found that while just 40% of the participants had actually seen a trans* patient, 78.5% were able to correctly answer the vignette questions for counseling a trans* patient provided by experts.⁴⁷ This study also revealed that though the majority of genetic counselors made the right decisions for the vignettes, nearly half felt less than completely confident about leading a session with a trans* individual.⁴⁷

2.4.1 Disclosure of trans* status

Disclosure of sex assigned at birth is vital when assessing individuals and families for genetic risk of certain diseases, such as those which are linked to the X-chromosome, or those which have sex-specific associated risks, such as breast cancer. However, it is important for rapport building, psychosocial support, and overall healthcare outcomes that this information is obtained in a culturally competent and sensitive manner.²⁵ Studies have shown that trans* individuals are generally okay with disclosing their gender identity to healthcare providers, but are more likely to feel comfortable doing so when they feel they are in an LGBT-friendly setting and are told why this information is necessary for a provider to have.⁴⁸ Surveys of trans* individuals found that

trans* participants reported that they would be far less likely to disclose their gender identity in a healthcare setting when they felt that it was not medically relevant.^{44,49} Disclosure of trans* status can be further complicated for trans* individuals who do not desire medical assistance in gender affirmation, where one study found that trans* individuals who did not want gender-affirming surgeries or hormone therapy were less likely to disclose their identities to providers.²⁴ Another contributing factor is that trans* individuals generally feel more comfortable disclosing this information when it is routinely collected from all patients,^{48,49} a consideration that has prompted some practices to ask for gender identity, birth sex, and legal sex on all intake forms.⁵⁰ One recent study even recommended that genetic counselors make it a habit to ask every client if anyone in the family has a gender identity which differs from their sex assigned at birth.⁵¹

Trans*-specific symbols (rather than simply a rainbow flag representing the entire LGBT community) in a clinic were also associated with a higher likelihood and comfort with disclosing trans* status.⁴⁴ A study which aimed to assess specific tools that genetic counselors could use to make disclosure of sex assigned at birth and gender identity easier for trans* patients found that providing a safe environment for disclosure and clarity regarding why the information was important was key.⁵¹ The trans* participants in this study gave concrete steps genetic counselors could take to achieve this, such as using validating language around gender identity and using inclusive pedigree symbology for trans* patients. Similarly, when asked how and when genetic counselors should ask about SO/GI, one study found that a sample of GLB individuals were okay with being asked as long as the inquiry was conducted in a “safe and appropriate way.”⁵² Though SO/GI can be sensitive information to elicit from patients, trans* perspectives are helpful to consider when determining how his information can be elicited in a genetic counseling session.

2.4.2 Pedigree nomenclature

A skill which is integral to the genetic counseling process is obtaining an accurate and well-documented family history. Counselors record this information in the form of a pedigree, a visual rendition of a family tree which makes familial health patterns and structures readily apparent to other genetic counselors and to other healthcare providers. In such diagrams, circles are used to represent female individuals in the family, and squares are used to represent male family members. There is currently no standardized symbology for trans* individuals that properly and sensitively documents sex assigned at birth as well as gender identity, both of which are important to obtain for aforementioned reasons.

One recent study involved gathering perspectives from trans* individuals on many aspects of the genetic counseling process including pedigree nomenclature. The study revealed that out of many options, trans* participants felt that the most appropriate was a symbol that aligned with their gender identity rather than their biological sex, with text underneath to clarify SAAB.⁴⁴ Though none of the participants of this study identified as nonbinary, the other trans* participants felt that a diamond shape with text to clarify SAAB would hypothetically be the most appropriate.⁴⁴ Another study by genetic counselors which included nonbinary individuals in their survey revealed similar findings—trans* individuals felt most comfortable with a symbol that best reflected their gender identity (a diamond for nonbinary individuals), with a note below regarding their SAAB.⁵¹ Though never before has the development of standard pedigree nomenclature been informed by the preferences of the population they are meant to represent, it is perhaps worth understanding and accommodating the preferences of trans* individuals on this issue due to the potential harm and loss of rapport that could be incurred in the event that a counselor uses the non-preferred symbology.⁵¹

Currently, no established standard pedigree nomenclature exists for all individuals of trans* identities. This lack of consensus of pedigree nomenclature can act as a systemic barrier for trans* individuals seeking genetic counseling, and can leave counselors feeling unprepared to provide counseling for this population.⁵³ The National Comprehensive Cancer Network (NCCN) has one set of guidelines for pedigree nomenclature for transgender men and women,⁵⁴ and the National Society of Genetic Counselors (NSGC) has recommendations that differ from those of the NCCN.⁵⁵ Neither recommendation is in line with what trans* people have recommended,⁴⁴ and suggestions from either of these entities regarding denotation of nonbinary trans* individuals are minimal. A recent survey of genetic counselors found that more than 75% did not know what symbol would be appropriate to use for trans* patients—of those who had a symbol that they would use, most (69.1%) cited the symbols recommended by NSGC, but 14.2% sided with the NCCN recommendations, or even had a symbol in mind that differed from both of these.⁵³ Though the clinical utility and functionality is untested, one study suggests that pedigree nomenclature should be reconceptualized, and proposes new symbology that clearly denotes sex and gender identity using astrological shapes.⁵⁶ Overall, studies on pedigree nomenclature for trans* individuals highlight the need for standardized and widely understood nomenclature in order to provide more competent care to trans* patients and to more easily communicate patients' trans* status to other genetic counselors and healthcare providers.

2.4.3 Language

Genetic counselors are trained to deliver information to patients in an accurate, sensitive, and easily understandable way with the ability to tailor information to the patient's level of understanding and specific needs. In all healthcare settings, trans* individuals have emphasized

the need for proper use of pronouns, gender-neutral language, and language that reflects a counselor's respect and understanding of trans* identities.⁵¹ It should be noted that trans* individuals have additionally provided their perspectives on culturally competent use of language in genetic counseling settings. When discussing medical conditions and options, studies have found that trans* individuals prefer specific, scientific language that is not gendered.^{24,44} For instance, a trans* individual may prefer that a provider use terms like breast cancer and ovarian cancer over gendered terms like women's cancers or female cancers. Additionally, trans* individuals have expressed discomfort over terms like male-bodied and female-bodied and tend to prefer gender neutral terms that refer specifically to the anatomical structure of interest, such as "people with uteruses."²⁴ Another study specifically within genetic counseling found that trans* people are more comfortable with counselors using chromosomal makeup rather than sex to describe sex modified risks—for instance, saying "you are at an increased risk because you have an X and Y chromosome" rather than "you are at an increased risk because you are male."⁵¹ Making these small changes can have a significant impact on the comfort of the genetic counseling client.

Several studies have shown that trans* individuals prefer and feel more at ease with counselors who understand and are comfortable speaking about the difference between gender and sex.^{44,51} Additionally, trans* individuals feel that it is important that providers know terminology and appropriate language surrounding trans* identities in general, with one study finding that individuals said they would be more likely to self-disclose their trans* status willingly and sooner to a provider who seemed like they knew about trans* issues.⁴⁴ One survey of genetic counselors found that while most genetic counselors felt comfortable using words like gender, transgender, and sexual orientation, counselors felt least confident in their knowledge of and ability to use words

like queer (39%), nonbinary/genderqueer (25.2%), cisgender (20.7%) and gender affirmation therapy (14.6%).⁴⁷ While genetic counselors often like to get first-person perspectives from patients on as many topics as possible, many trans* individuals express frustration or exhaustion over having to explain their gender identity to providers,⁵ and having culturally competent and appropriate understandings of common terms within the trans* community can assist in rapport building and overall satisfaction.⁵¹

2.4.4 Risk assessment

There are genetic counseling situations commonly encountered where the biological sex of the patient and their family members is relevant to risk assessment for certain genetic conditions. While many conditions are modified by sex and others are directly inherited on sex chromosomes, the most well-studied with regard to trans* counseling issues are hereditary breast and ovarian cancer syndromes. Like other key components of genetic counseling, risk assessment for trans* patients can be complicated when SAAB is not elicited, and when records of anatomical structures are not properly recorded in medical records for trans* patients.⁵⁷ Counseling for these cancer predisposing syndromes is additionally complicated by differing motivations for testing, past gender-affirming surgeries, and the decision to pursue hormone replacement therapy (HRT).^{45,47} Unique motivations for assessing cancer risk may be directly related to a person's gender identity.⁴⁷ A genetic counseling case study published on high risk BRCA testing for three trans* individuals highlighted unique motivations for testing, where transgender men and transmasculine individuals may pursue testing at younger ages in an attempt to obtain coverage for "top surgery" and the more invasive prophylactic mastectomy, and transgender women and transfeminine individuals may pursue testing in order to have more information on risks of HRT.⁵⁸

Though data are still preliminary, one large study in 2013 found that breast cancer incidence in transgender women on estrogen supplements was not significantly elevated above the incidence in the general population of individuals assigned male at birth.⁵⁹ Additionally, the difference in risk for BRCA mutation carriers to develop breast cancer appears not to be significantly different between AMAB trans* individuals using estrogen and males not using estrogen.⁶⁰ However, other studies report that as the body of research accumulates, some data suggest an increased lifetime risk for breast cancer in trans* individuals correlated with the duration of estrogen use.⁶¹ One paper published in 2017 attempted to summarize what was currently known in the literature regarding high-risk breast cancer in trans* individuals and provided recommendations for clinical care and surveillance.⁶⁰ This group recommended that AFAB trans* individuals with BRCA mutations who undergo risk reducing bilateral mastectomies be surveilled the same as a cisgender woman with the same surgery and mutation would be according to NCCN guidelines.⁵⁰ All recommendations take into account the physical and mental wellbeing of a trans* patient. For instance, in the event that a trans* individual using gender-affirming estrogen is diagnosed with breast cancer, a discussion about stopping estrogen therapy is relevant, but providers should keep in mind the health benefits of gender affirmation and the mental health of the patient.⁶⁰ Despite gaps in knowledge due to a lack of data in risk assessment for trans* individuals, accurate risk assessment is critical to the genetic counseling process,⁴⁷ and communication about what is currently unknown is a familiar skill to a practiced genetic counselor.

2.4.5 Psychosocial considerations

Psychosocial care of the client is integral to the genetic counseling process. Many psychosocial strategies commonly used in counseling sessions would be appropriate to use in

counseling trans* patients. However, research has identified additional psychosocial counseling considerations for counseling trans* patients. Though counselors should be well aware that a person's trans* status is not indicative of a mental illness, it's important to be aware that trans* populations report higher rates of anxiety and depression, which often stem from coping with social stigma.⁴⁰ Trans* individuals are more likely to experience a lack of familial support and may have non-traditional familial structures or "chosen" families, which may make topics such as family history of disease a sensitive topic.⁵¹ Though assessing a client's support network is a psychosocial counseling strategy which is important to all clients, this tool can be especially important for trans* patients, as increased social support has been linked to better coping skills, lower risks of anxiety and depression, and overall better healthcare outcomes for trans* patients.⁶² Proper use of pronouns and validation of a trans* individual's identity during a healthcare encounter has additionally been found to be vital to a trans* individual's psychosocial care.⁵¹ While validation and recognition of a client's mental state is vital to every counseling session, these skills become even more important for a population who may be more accustomed to or fearful of dismissal or discrimination in a healthcare setting.

As mentioned previously, it is unclear whether use of estrogen for hormone replacement therapy could increase the risk of developing breast cancer in AMAB trans* individuals. Despite this unknown, it is important to provide the most accurate and up to date information on these risks without posing a barrier to a person's goals for gender affirmation, and these risks should be weighed against the psychosocial benefit of receiving this gender-affirming care.^{60,61} Moreover, it is important that in all healthcare encounters with trans* clients individuals be allowed to freely share their experiences and have those experiences validated and taken into account by their healthcare professionals.⁴⁰ Studies have shown that trans* individuals who do not desire medical

services, such as HRT or gender-affirming surgeries, are less likely to share their trans* status with their providers for fear that they will attempt to push those services on them.²⁴ In this and many other scenarios, there is the risk of causing psychosocial harm when making incorrect assumptions, in this case the assumption that all trans* individuals desire some type of medical intervention for gender affirming care. Genetic counselors are trained in strategies to create environments that are safe for emotional expression and self-disclosure—these strategies would be particularly important for trans* patients, especially with issues surrounding personal experience and trans* status.

2.5 Addressing the problem: Creating educational resource materials for genetic counseling training programs

Multiple studies have found an overwhelming desire from genetic counselors for more education, both during their time in school and post-graduation, on counseling trans* and LGBT individuals.^{46,47,53,63} Education regarding trans* health in professional school settings has shown to positively influence providers' confidence in their ability to provide culturally competent service and to improve outcomes for trans* patients.⁶⁴⁻⁶⁶ The Accreditation Council for Genetic Counseling's (ACGC) Standards of Accreditation for Graduate Programs in Genetic Counseling lists multicultural competency as a required component of every genetic counselors' psychosocial education.⁶⁷ Though no mention is made specifically that counseling for gender and sexual minorities is a required component of every genetic counselors' education, the use of educational resource material on genetic counseling for trans* individuals would likely assist program directors in satisfying this accreditation requirement of their students' psychosocial capabilities. The ACGC additionally provides to counselors and educational programs a list of practice-based

competencies that all genetic counselors should continually work on and strive towards during their educational training.⁶⁸ An understanding of one's own biases as a means to providing culturally competent care is one of the abilities that ACGC deems critical. In addition, competency in constructing relevant and appropriate pedigrees, knowledge of reproductive and preventative care, and appropriate and accurate assessment of risk are integral competencies to genetic counseling.⁶⁸ As discussed, trans* identities complicate and add nuance to these daily tasks that a genetic counselor must accurately and sensitively complete. Education on trans* health topics and genetic counseling for trans* individuals would support current genetic counselor training to more successfully employ these competencies for all patients of all gender presentations and identities.

2.5.1 Developing educational resource materials

Multicultural competency has long been integral to the goals of genetic counseling training programs. One early paper outlines the goals of multicultural genetic counseling education as being “knowledge of relevant ethnocultural groups, ethnocultural self-awareness, and an understanding of institutional and social barriers to services.”⁶⁹ In order to achieve mastery of these three tenets of multicultural genetic counseling, educators in genetic counseling programs have used a variety of resource materials. Many of these educational strategies and materials can easily be adapted for trans* genetic counseling education. A 1993 paper suggested that current genetic counselors should have a large role in developing the multicultural genetic counseling educational materials needed to educate future counselors.⁷⁰ Therefore, any new materials made should draw upon perspectives from and research already done by practicing genetic counselors, specifically genetic counselors who have experience in education.

Genetic counseling programs have traditionally used multiple forms of educational tools and assessment in order to cater to multiple learning styles. Commonly used educational resource material in genetic counseling programs include textbook readings, curated primary literature readings, presentation slides, relevant videos and demonstrations, roleplaying activities, and written/verbal assessments. Much of the research on genetic counseling education emphasizes the importance of learning through practice and use of multiple kinds of educational tools for different learning styles.¹⁰ Several of these curricular materials for trans* counseling can be made to allow students to learn, discuss and practice learned skills.⁷⁰

3.0 Manuscript

3.1 Background

3.1.1 Gender and trans* identities

Language surrounding sexual orientation and gender identity is constantly evolving and changing in order to meet the needs of sexual and gender minorities. While a person who is cisgender identifies with the gender most often ascribed to the sex they were assigned at birth, a person is considered to be transgender when their gender identity is incongruent with or socially perceived to be different from the sex they were assigned at birth (SAAB). In 2016, it was estimated that 1.4 million adults in the United States identified as transgender, which does not account for transgender youth, and may underestimate those who identify as gender non-binary.² Despite the nebulous nature of this terminology, a shared language on these topics between providers and patients is vital for successful communication, provision of healthcare, and information gathering.¹¹

Though many transgender identities exist outside of binary understandings of gender, generally someone who was assigned male at birth (AMAB) who identifies as a woman is considered a transgender woman, often denoted as being male to female (MtF). Likewise, someone who was assigned female at birth (AFAB) who identifies as male would be a transgender man, or female to male (FtM). Trans* is a term that has emerged in recent years to describe all individuals who do not identify as a cisgender man or cisgender woman. Though transgender and trans have also been used as blanket terms for this population in the past, the designation trans* provides

more inclusivity of identities that are outside of a binary understanding of gender, resulting in a term for a community that is less medicalized and more inclusive of many individuals' understanding of their own gender.^{18,19}

3.1.2 Healthcare disparities in trans* populations

Multiple organizations such as the American Public Health Association and the 2020 Healthy People campaign have recognized the need for improvement regarding the health disparities trans* people in America face.^{3,4} LGBT (Lesbian, Gay, Bisexual, and Transgender) populations experience higher rates of substance use, tobacco use, and obesity than the population average,⁴ and mental health concerns such as depression and anxiety appear to be elevated in the trans* population.⁵ A national survey found that 41% of trans* individuals had attempted suicide—four times the national average.⁵ Trans* individuals are more likely to experience assault and intimate partner violence than cisgender individuals, and are much more likely to delay important medical treatment out of fear of discrimination.⁶ In addition, trans* people are more likely to be uninsured, and disproportionately experience unemployment, homelessness, legal discrimination in adoption and marriage, and loss of support from parents or family members, all of which are social determinants of health.⁵

Healthy People 2020, a national initiative focused on using scientific data to set goals for improving the health of Americans, cited a lack of knowledgeable providers for LGBT individuals as one of the top barriers negatively affecting the health of trans* people.⁴ Nearly a quarter of trans* individuals report having been refused care, and over a quarter had experienced harassment in a healthcare setting.⁵ Though studies suggest that knowledge of both sex and gender identity is vital for a provider to have in order to deliver appropriate care,¹¹ a trans* person is more likely to

experience discrimination from a healthcare provider when their trans* status is known to them.⁵ This suggests that disclosing one's gender identity will improve a person's care only if the provider receiving the disclosure is educated on trans* health and respectful towards trans* identities.⁷ Overall, of those who had seen a healthcare provider in the last year, one-third of trans* people reported at least one experience of harassment or discrimination with their healthcare provider.⁶

3.1.3 Trans* healthcare guidelines and best practices

Increasingly, educational resource material has become available to practicing clinicians and clinical training programs for the appropriate treatment of trans* patients. Many professional institutions and advocacy organizations have released trainings, guidelines, and best practices for treating trans* patients and for educating young professionals about trans* healthcare.^{16,25,28-35} In 2010, the American Counseling Association released their Competencies for Counseling with Transgender Clients,²⁸ and in 2015, the American Psychological Association released Guidelines for Psychological Practice with Transgender and Gender Nonconforming People.¹⁶ These documents outline suggested competencies for all counselors to achieve in order to appropriately provide care to trans* patients. Other professional health organizations that have released their own recommendations or guidelines for working with trans* patients.

Trans* advocacy organizations play an important role in informing the public of trans* issues, as well as their history. Many groups have published guidelines or recommendations for healthcare providers on how to best serve the population. The Center of Excellence for Transgender Health at the University of California San Francisco has created coursework for medical professionals including educational videos, perspectives from transgender individuals, and the opportunity for self-assessment of learned skills in providing care for transgender

individuals in the form of case studies.³⁴ The National Center for Transgender Equality has also created resources not only for providers, but for trans* individuals navigating the medical system as well.³³ The World Professional Association for Transgender Health (WPATH) has also released their own standards of care for the health of trans* individuals.³⁵

Research within trans* populations has allowed individuals seeking to provide competent care a look into the opinions from those who are experts on the topic—trans* individuals themselves. Through these studies, trans* individuals have been able to communicate recommendations to healthcare providers about how they can provide them with the best care possible.^{21,39-42} Trans* individuals have voiced their opinions on best practices for a trans*-friendly office environment, appropriate ways to elicit gender and sex information, and actions that make a difference between a positive and a negative healthcare encounter.

Despite the knowledge we have about trans* healthcare, gaps in education and research continue to affect the treatment trans* people receive from healthcare providers. Studies have shown reluctance from LGBT individuals to participate in research due to surveys and paperwork lacking in inclusivity, which may cause these unknowns to persist in clinical research.¹⁵ Despite the publication of clinical guidelines in many specialty areas, a recent study found that up to a third of all medical schools currently have no hours of LGBT health curriculum.⁴³ This, in addition to the fact that many standard educational materials lump healthcare for trans* individuals in with the education needed to serve LGB individuals without addressing issues specific to gender minorities,²¹ demonstrates that the vast majority of clinical healthcare providers may not have relevant information to provide appropriate trans* healthcare despite its increased availability. Overall, the knowledge we have is incomplete, and what we do have may not be reaching providers in educational settings.

3.1.4 Genetic counseling for trans* individuals

Genetic counseling is defined by the National Society of Genetic Counselors (NSGC) as “the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.”⁸ While many efforts have been made to promote and teach multicultural competency in genetic counseling, like all other forms of healthcare in the United States, inequities in access to genetic counseling and quality of care persist for minority populations.⁹ Despite gaps in knowledge, some studies have helped to identify best practices for genetic counselors to utilize in clinical situations with trans* clients.

Disclosure of sex assigned at birth is vital when assessing individuals and families for genetic risk of certain diseases, such as those which are linked to the X-chromosome, or those which have sex-specific associated risks, such as breast cancer. However, it is important for rapport building, psychosocial support, and overall healthcare outcomes that this information is obtained in a culturally competent and sensitive manner.²⁵ Studies have shown that trans* individuals are generally willing to disclose their gender identity to healthcare providers, but are more likely to feel comfortable doing so when they feel they are in an LGBT-friendly setting and are told why this information is necessary.⁴⁸ Surveys of trans* individuals found that they would be far less likely to disclose their gender identity in a healthcare setting when they felt that it was not medically relevant,^{44,49,51} or when they do not desire medical assistance in gender affirmation.²⁴ Another contributing factor is that trans* individuals generally feel more comfortable disclosing this information when it is routinely collected from all patients,^{48,49} a consideration that has prompted some practices to ask for gender identity, birth sex, and legal sex on all intake forms.⁵⁰ Trans*-specific symbols (rather than symbols such as a rainbow flag representing the entire LGBT

community) in a clinic were also associated with a higher likelihood and comfort with disclosing trans* status.⁴⁴

A skill which is integral to the genetic counseling process is obtaining an accurate and well-documented family history in the form of a pedigree. In such diagrams, circles are used to represent female individuals in the family, and squares are used to represent male family members. There is currently no standardized symbology for trans* individuals that properly and sensitively documents sex assigned at birth as well as gender identity, both of which are important to obtain. The National Comprehensive Cancer Network (NCCN) has guidelines for pedigree nomenclature for trans* individuals,⁵⁴ and the National Society of Genetic Counselors (NSGC) has recommendations that differ from those of the NCCN.⁵⁵ Neither recommendation is in line with what trans* people have recommended,⁴⁴ and suggestions from either of these entities regarding denotation of nonbinary trans* individuals are minimal. A recent survey of genetic counselors found that more than 75% did not know what symbol would be appropriate to use for trans* patients.⁵³ Trans* study participants felt that the most appropriate was a symbol that aligned with their gender identity rather than their biological sex, with text underneath to clarify SAAB.⁴⁴ Though never before has the development of standard pedigree nomenclature been informed by the preferences of the population they are meant to represent, it is perhaps worth accommodating the preferences of trans* individuals on this issue due to the potential loss of rapport that could be incurred in the event that a counselor uses symbols that are viewed as disrespectful.⁵¹

. In all healthcare settings, trans* individuals have emphasized the need for proper use of pronouns, gender-neutral language, and language that reflects a counselor's respect and understanding of trans* identities.⁵¹ Trans* individuals have provided their perspectives on culturally competent use of language in genetic counseling settings in several studies. When

discussing medical conditions and options, trans* individuals prefer specific, scientific language that is not gendered.^{24,44} Additionally, trans* individuals prefer gender neutral terms that refer specifically to anatomical structures, such as “people with uteruses.”²⁴ Another study found that trans* people are more comfortable with counselors using chromosomal makeup rather than sex to describe sex modified risks—for instance, saying “you are at an increased risk because you have an X and Y chromosome” rather than “you are at an increased risk because you are male.”⁵¹ Trans* individuals prefer and feel more at ease with counselors who are comfortable speaking about the difference between gender and sex.^{44,51} One survey of genetic counselors found that while most genetic counselors felt comfortable using words like gender, transgender, and sexual orientation, counselors felt least confident in their knowledge of and ability to use words like queer (39%), nonbinary/genderqueer (25.2%), cisgender (20.7%) and gender affirmation therapy (14.6%).⁴⁷ While genetic counselors often like to get first-person perspectives from patients on as many topics as possible, many trans* individuals express frustration or exhaustion over having to explain their gender identity to providers,⁵ and having appropriate understandings of common terms within the trans* community can assist in rapport building and overall satisfaction.⁵¹

There are commonly encountered indications in genetic counseling where the biological sex of the patient and their family members is relevant to risk assessment. Like other key components of genetic counseling, risk assessment for trans* patients can be complicated when SAAB is not elicited, and when records of anatomical structures are not properly recorded in medical records for trans* patients.⁵⁷ Counseling for cancer predisposing syndromes is complicated by differing motivations for testing, past gender-affirming surgeries, and the decision to pursue hormone replacement therapy (HRT).^{45,47} Unique motivations for assessing cancer risk may be directly related to a person’s gender identity.⁴⁷ A genetic counseling case study published

on high risk BRCA testing for three trans* individuals highlighted unique motivations for testing, where transgender men and transmasculine individuals may pursue testing at younger ages in an attempt to obtain coverage for “top surgery” and the more invasive prophylactic mastectomy, and transgender women and transfeminine individuals may pursue testing in order to have more information on risks of HRT.⁵⁸ Decision making based on genetic testing results should take into account the physical and mental wellbeing of a trans* patient.⁵⁰ For instance, in the event that a trans* individual using gender-affirming estrogen is diagnosed with breast cancer, a discussion about stopping estrogen therapy is relevant, but providers should keep in mind the health benefits of gender affirmation and the mental health of the patient.^{60,61}

Psychosocial care of the client is integral to the genetic counseling process, and there are considerations to take into account for trans* clients. Though counselors should be well aware that a person’s trans* status is not indicative of a mental illness, it’s important to be aware that trans* populations report higher rates of anxiety and depression, which often stems from coping with social stigma.⁴⁰ Trans* individuals are more likely to experience a lack of familial support and may have non-traditional familial structures or “chosen” families, which may make topics such as family history of disease a sensitive topic.⁵¹ Assessing a trans* client’s support network is an important psychosocial tool, as increased social support has been linked to better coping skills, lower risks of anxiety and depression, and overall better healthcare outcomes for trans* patients.⁶² Proper use of pronouns and validation of a trans* individual’s identity during a healthcare encounter has additionally been found to be vital to a trans* individual’s psychosocial care.⁵¹ It is important that in all healthcare encounters with trans* clients individuals be allowed to freely share their experiences and have those experiences validated and taken into account by their healthcare

professionals.⁴⁰ As always, it is important to not make assumptions about clients and allow for open dialogue. Genetic counselors should use psychosocial skills and non-directive counseling.

3.1.5 Development of educational resource materials

Multiple studies have found an overwhelming desire from genetic counselors for more education, both during their time in school and post-graduation, on counseling trans* and LGBT individuals.^{46,47,53,63} Education regarding trans* health in professional school settings has shown to positively influence providers' confidence in their ability to provide culturally competent service and to improve outcomes for trans* patients.⁶⁴⁻⁶⁶ The Accreditation Council for Genetic Counseling's (ACGC) Standards of Accreditation for Graduate Programs in Genetic Counseling lists multicultural competency as a required component of every genetic counselors' psychosocial education.⁶⁷ Though no mention is made specifically that counseling for gender and sexual minorities is a required component of every genetic counselors' education, the use of educational resource material on genetic counseling for trans* individuals would likely assist program directors in satisfying this accreditation requirement of their students' psychosocial capabilities. The ACGC additionally provides to counselors and educational programs a list of practice-based competencies that all genetic counselors should continually work on and strive towards during their educational training.⁶⁸ As discussed, trans* identities complicate and add nuance to these daily tasks that a genetic counselor must accurately and sensitively complete.

Genetic counseling programs have traditionally used multiple forms of educational tools and assessment in order to cater to multiple learning styles. Commonly used educational resource material in genetic counseling programs include textbook readings, curated primary literature readings, presentation slides, relevant videos and demonstrations, roleplaying activities, and

written/verbal assessments.¹⁰ Several of these curricular materials for trans* counseling can be made to allow students to learn, discuss and practice learned skills.⁷⁰

3.1.6 Purpose and specific aims

The aim of this study was to design and distribute a Qualtrics survey to ACGC-accredited genetic counseling programs to assess current curricula in genetic counseling for transgender and gender nonconforming (GNC) individuals, as well as programs' perception of educational resource material on this topic. An additional aim was to use the survey data, primary literature involving perspectives from transgender and GNC individuals, and best practices guidelines from other healthcare professions and LGBT health organizations to develop and distribute educational resource materials for genetic counseling programs. The purpose of this study was to make educational resources on genetic counseling for trans* individuals accessible to programs with the intention that more genetic counseling programs may include trans* issues in genetic counseling as part of their genetic counseling program curriculum.

3.2 Materials and Methods

3.2.1 Survey design

A survey was designed to ascertain educational content regarding genetic counseling for transgender and gender non-conforming individuals in ACGC accredited programs. The introductory survey text outlined several important terms about sexuality and gender that were

important to know for the purpose of completing the survey, and stated that the survey was optional, there were no benefits to participating, minimal risk was associated with participation, information was collected anonymously, and there was no compensation for participating. This study was determined by the IRB as not human research (Appendix D). The design of the survey was aided by a prior thesis project that elicited curriculum content related to palliative care of ACGC programs.⁷¹

The survey consisted of 21 questions divided into 4 sections and was developed using the Qualtrics survey platform. The first section assessed the participant's affiliation with an ACGC-accredited program. If the participant was not, the survey used skip logic to end the survey. The second section focused on how a program currently addressed education on genetic counseling for trans* individuals by asking about specific types of programming used or what barriers prevented exploration of this topic. The third section of the survey elicited whether the participant felt that having educational resource materials on genetic counseling for trans* individuals would be beneficial, and if so, what resource material was most desired. The fourth section assessed program demographics. A copy of the survey text can be found in Appendix B.

3.2.2 Participants

The population of interest for this study was program leadership of ACGC-accredited genetic counseling programs. Program leadership is defined by ACGC as university employees with “designated time that is free from clinical service, research efforts, and institutional responsibilities to perform their educational and administrative duties directly related to the genetic counseling program.”⁶⁷ Program leadership includes head instructors and faculty employed by the university who oversee program educational standards and curriculum as well as ACGC

accreditation status. These members of program leadership are generally members of or connected to the Association of Genetic Counseling Program Directors. For this reason, participants were recruited through this organization via a listserv email. The introductory survey text emphasized that just one member of program leadership should participate per program

The recruitment email included a brief introduction to the study, including a link to participate and contact information for the lead researcher (Appendix A). Participants accessed the survey through a single web link. The survey was open for participation between October 1st, 2020 and December 15th, 2020. During this time, two brief reminder emails were sent on October 23rd, 2020 and November 13th, 2020.

3.2.3 Data collection and analysis

Data was collected anonymously through the Qualtrics system. Microsoft Excel was used to perform descriptive statistics on the data collected through Qualtrics. The survey received twelve partial responses (less than 50% of the survey was completed), which were excluded from analysis.

3.2.4 Creation of educational resource material

One goal of the survey was to assess which kinds of educational resource materials would be most beneficial for program leadership. The types of educational materials created are based on the results of the survey, prioritizing those materials the respondents felt would be most helpful. Additionally, due to the lack of interest in more didactic learning materials, an overview sheet of trans* genetic counseling issues with strategies was made to provide valuable background in a

concise manner. All educational resource materials were developed using Microsoft Word and were informed by existing guidelines, research, and best practices in trans* healthcare.

Clinical case scenarios were created to emulate issues specific to trans* genetic counseling that had been documented in the literature. Lists of resources were pulled from the literature review done for this project. Clinical case examples/roleplay opportunities were created for the three most common genetic counseling specialties—cancer genetics, prenatal genetics, and pediatric genetics, to give examples of trans*-specific counseling

3.3 Results

3.3.1 Participant demographics

Twenty ACGC-accredited genetic counseling programs responded to the survey. At the time the survey was distributed, fifty-six programs held ACGC accreditation, making the response rate 35.7% (20/56). Partial responses were not included in the analysis. No responses from programs not accredited by the ACGC were received. Summary of demographic data can be found in Table 1. All six regions of North America under ACGC accreditation were represented in the data, with the largest representation at 40% (8/20) from region 4 (AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI, Ontario). The majority of respondents (75%) were from programs that have been in existence for more than 10 years. Half (50%) of all respondents were affiliated with programs located in a major city, and the majority (75%) were programs that admitted 5-10 students a year.

Table 1: Participant Demographics

Variable	Number of programs (n = 20)
Region	
Region 1 (CT, MA, ME, NH, RI, VT, CN Maritime Provinces)	1 (5%)
Region 2 (DC, DE, MD, NJ, NY, PA, VA, WV, PR, VI, Quebec)	3 (15%)
Region 3 (AL, FL, GA, KY, LA, MS, NC, SC, TN)	3 (15%)
Region 4 (AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI, Ontario)	8 (40%)
Region 5 (AZ, CO, MT, NM, TX, UT WY, Alberta, Manitoba, Saskatchewan)	3 (15%)
Region 6 (AK, CA, HI, ID NV, OR, WA, British Columbia)	2 (10%)
Age of Program	
0-3 years	4 (20%)
4-9 years	1 (5%)
>10 years	15 (75%)
Size of Program	
2-4 students	0 (0%)
5-10 students	15 (75%)
11-18 students	4 (20%)
18+ students	1 (5%)
Environment/Location of Program	
Major city (Population of at least 800,000)	10 (50%)
Large city (Population 500,000 to 800,000)	4 (20%)
Medium city (Population 100,000 to 500,000)	5 (25%)
Small city (Population 50,000 to 100,000)	1 (5%)
Town/Rural Environment (Population 50,000 or less)	0 (0%)

3.3.2 Current trans* genetic counseling education in coursework/curricula

While 90% of participating counseling programs reported some training regarding genetic counseling involving lesbian, gay, and bisexual individuals, fewer reported training specific to trans* populations (80%).

All programs currently integrating education for counseling trans* clients into their curriculum (n=16) state that the primary reason this education is implemented is because the program's faculty feel it is important. Additionally, the majority of programs present their trans* genetic counseling education across both years of the master's degree program (56.3%). See **Appendix E** for survey data.

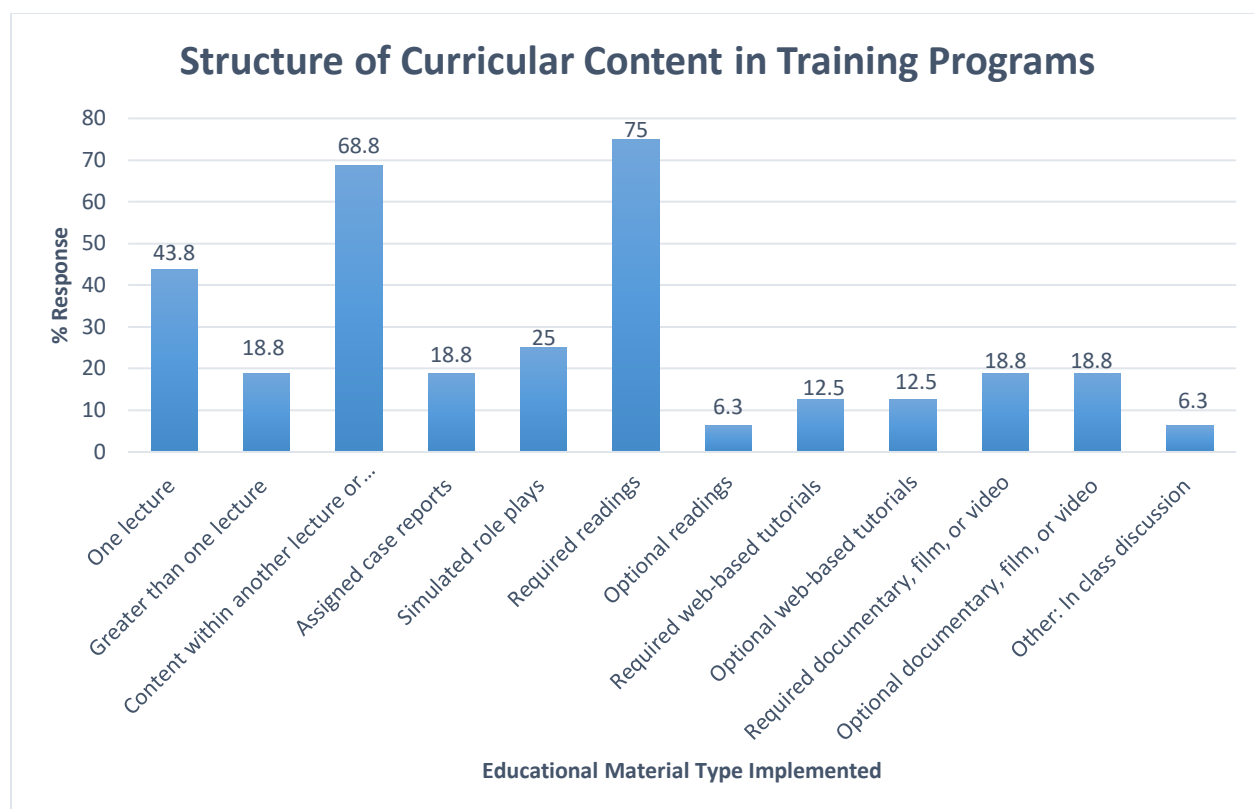


Figure 1: Percent response of types of curricular content on trans* genetic counseling issues within genetic counseling training programs. N=16

Figure 1 summarizes the educational methodologies implemented by those programs with education on trans* genetic counseling. Of those programs implementing educational resource material on genetic counseling for trans* individuals, the most common educational methodologies include required readings (75%) and integrating trans* healthcare into lectures on other counseling topics (68.8%). One respondent cited using in-class discussion as a tool utilized in their program to educate students on counseling trans* individuals.

Table 2: Instructors for Trans* Genetic Counseling Curriculum

Instructor(s) for Trans* Genetic Counseling Curriculum	Number of Programs N=16
Healthcare provider other than a genetic counselor with experience in caring for transgender/gender nonconforming individuals	6 (37.5%)
Genetic counselor	14 (87.5%)
Individual from a transgender health advocacy group	2 (12.5%)
Other instructor, please explain: MSSW social worker faculty member of GC program	1 (6.3%)

Table 2 summarizes the instructors of trans* genetic counseling content in programs with trans* genetic counseling education. The majority (87.5%) of programs with trans* health education within their curriculum reported that a genetic counselor delivered the educational materials on trans* counseling. Two programs involved education from an individual from within a transgender health advocacy group, and six involved a healthcare provider, other than a genetic counselor, with experience in caring for trans* clients.

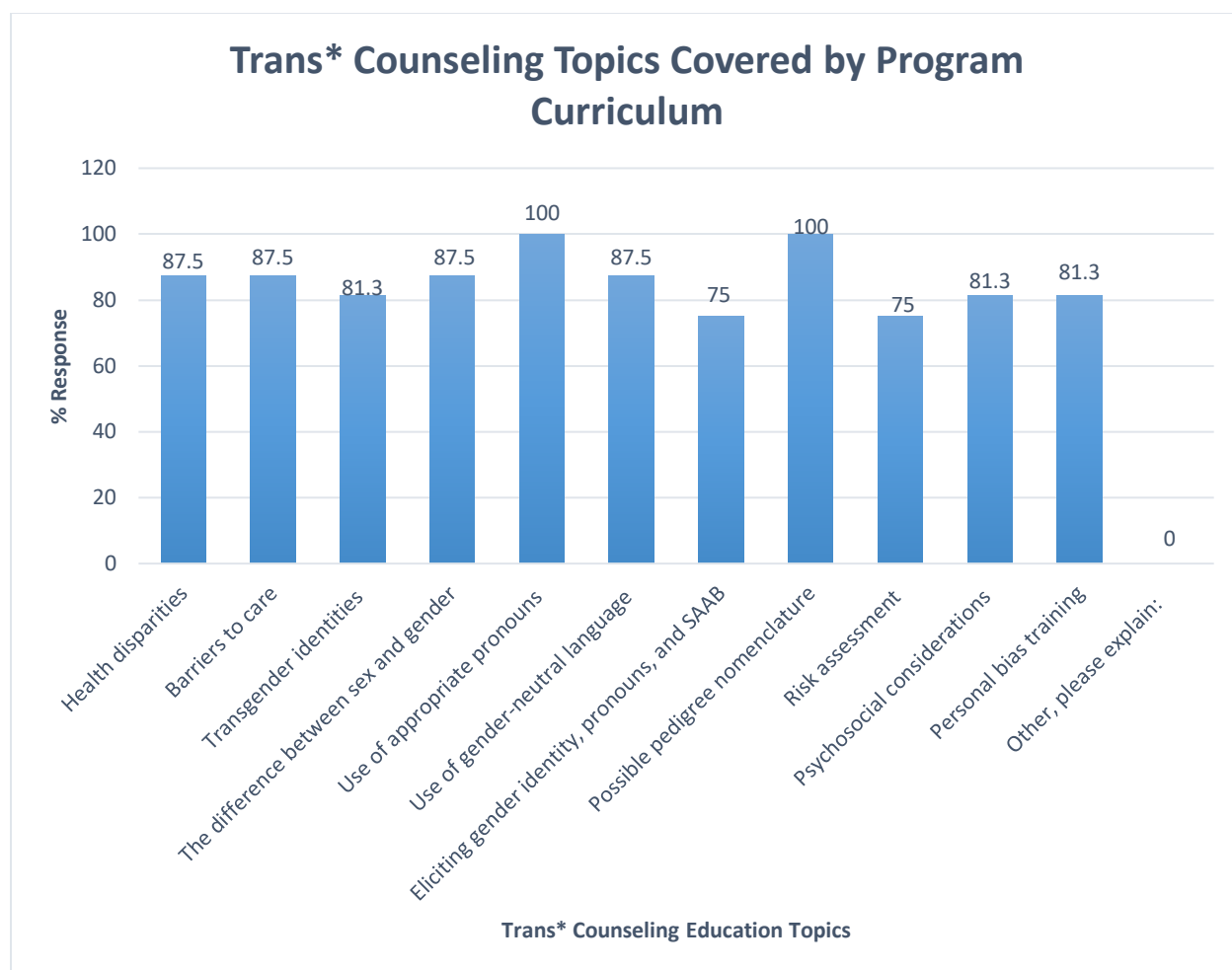


Figure 2: Percent response of trans* genetic counseling topics included in program curriculum. N=16

Figure 2 summarizes topics covered by programs as part of their trans* genetic counseling education. Responding ACGC-accredited genetic counseling programs with trans* genetic counseling education included a variety of key topics in their curriculum. All respondents covered appropriate language and possible pedigree nomenclature when working with trans* clients. The least covered topics were eliciting gender identity, pronouns, and sex assigned at birth (75%) as well as risk assessment for trans* patients (75%).

Of those programs not currently employing any education on genetic counseling for trans* populations (n=4), all of them cited a lack of appropriate educational materials to be one barrier to

providing this education to their students. Two out of four respondents cited this lack of appropriate educational materials to be the most important barrier. Of note, no programs suggested the lack of education was due to disinterest from students or an underlying belief that the education was unnecessary or irrelevant to genetic counseling (Table 3).

Table 3: Barriers to Implementing Trans* Genetic Counseling Education

Reason for Lacking Education on Trans* Genetic Counseling	Number of Programs (n=4) (Choose all that apply)
Lack of knowledgeable faculty/staff	2
Lack of available time in the program to cover this material	1
Lack of appropriate educational materials	4
Students are disinterested	0
The education is unnecessary/irrelevant to genetic counseling training	0
Other, please describe: Until the last couple of years it just never occurred to include specific education on this with exception of occasional examples included in scenarios discussed in class. We're at a point that we want to include direct instruction on this but want to be sure to do it in a sensitive and appropriate way.	1

Respondents were additionally asked questions regarding their feelings on issues pertaining to genetic counseling for trans* populations. When asked, “Should ACGC require that all genetic counseling students acquire the ability to counsel gender diverse populations?”, half of the respondents (10/20) answered “definitely yes” or “probably yes,” 20% (4/20) responded “probably not,” and 30% were unsure (Appendix E). Notably, 95% (19/20) of respondents either agreed or strongly agreed with the statement “Genetic counseling programs should incorporate education on counseling transgender/gender nonconforming individuals into their curriculum.” One respondent neither agreed nor disagreed (Appendix E).

3.3.3 Perceived benefits of educational resources for trans* genetic counseling

The majority of respondents felt that their program would benefit from access to educational resource materials (85%). 10% (2/20) responded “unsure/maybe” as to whether these materials would be beneficial. One respondent did not feel these resources would be beneficial, citing, their program had a “fairly robust” curriculum related to LGBT and trans* issues in genetic counseling, and therefore access to these materials would likely not significantly influence the education already in place for their students. Those who responded either “yes” or “unsure/maybe” to the question of whether educational resource materials on trans* genetic counseling would be helpful were then prompted to select from a menu or provide their own ideas of which educational resource materials would be most helpful for them to have. Figure 3 summarizes which educational materials were perceived to be most beneficial to programs. Three respondents typed in their own responses to this question, citing speaker’s bureaus, expert lecturers, and a video series from genetic counselors as potentially helpful resources.

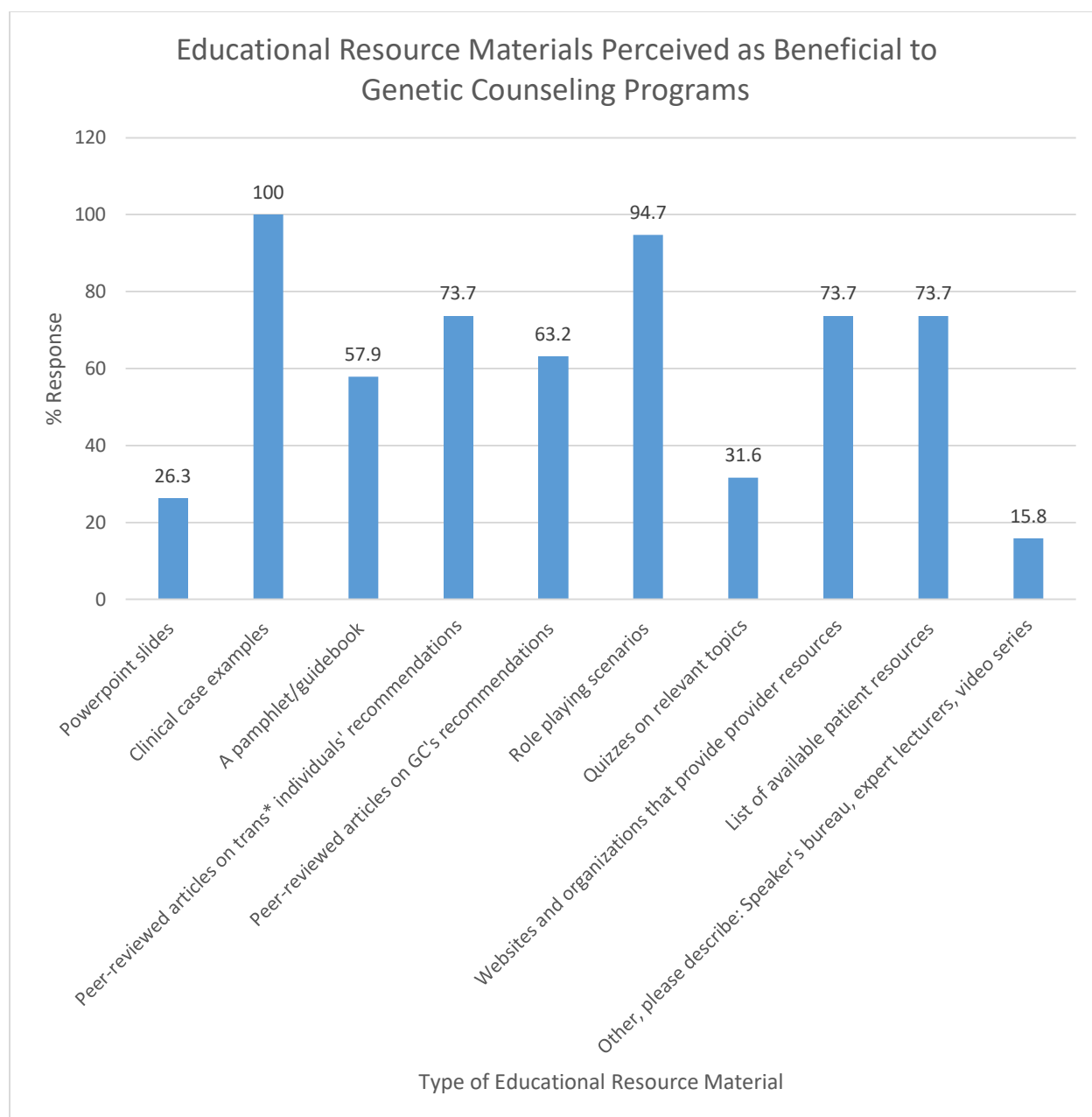


Figure 3: Percent response of educational resource materials perceived to be beneficial to genetic counseling programs (N=19)

The educational resource materials developed for this study were chosen based upon the responses to this question, which are depicted in Figure 3. Those resources that were perceived to be most helpful were clinical case examples or vignettes that highlight trans*-specific

considerations in genetic counseling (100%), role playing scenarios that can be used to practice counseling skills (94.7%), a list of websites and organizations that provide valuable resources for working with trans* clients (73.7%), a list of available patient resources for trans* clients (73.7%), a list of peer-reviewed articles on trans* individuals' recommendations to genetic counselors (73.7%), and a list of peer-reviewed articles on genetic counselors' recommendations on working with trans* individuals (63.2%). Due to the significant amount of interest for all of the educational resource materials listed in the survey, those materials with greater than 60% interest were created. Because data on genetic counseling for trans* individuals are not yet widely available, the list of peer-reviewed articles on trans* individuals' recommendations to genetic counselors and the list of peer-reviewed articles on genetic counselors' recommendations on working with trans* individuals have been combined. Additionally, the case examples were combined with roleplay opportunities based on the clinical scenarios. These resources will be provided to programs via the Association of Genetic Counseling Program Directors (Appendix C).

3.4 Discussion

Survey responses reveal novel information about how genetic counseling training programs educate students about providing care for trans* individuals. This study found that most genetic counseling programs currently incorporate education on trans* counseling issues into their program curriculum. This material is generally presented to students across both years of the master's program, and there are a variety of educational resource materials used and topics covered. Though the majority of programs have some degree of education on trans*-specific

counseling issues, almost all programs desire access to more educational resource materials specifically on genetic counseling for trans* clients.

3.4.1 Trans* genetic counseling education

Most respondents to this survey were from programs older than 10 years (75%), located in a major city with a population of at least 800,000 (50%), and admit class sizes of students within the 5-10 range (75%). According to the ACGC website's list of currently accredited programs, about 25% of programs are currently housed in urban settings with populations greater than 800,000.⁷² Additionally, 67% of the currently accredited genetic counseling programs have been in existence for at least four years.⁷² The average size of ACGC-accredited programs falls within the 5-10 students per class range. This survey revealed that the majority of genetic counseling programs (80%) currently incorporate education on genetic counseling for trans* individuals into their curriculum. Older, more established programs appear to be overrepresented in the sample captured by this survey. Newer programs have more recently developed program curriculum, which may be more encompassing of emerging health issues facing marginalized groups such as trans* individuals.

Most genetic counseling programs cover trans* health topics already, which is in line with the literature that suggests that trans* healthcare concerns are becoming increasingly important to and prioritized in other healthcare professions.^{3,16,28,29,36} This is unsurprising, given the emphasis on psychosocial care for the client that is built into genetic counseling program curriculum.⁶⁷

Topics covered by these programs align with those topics that previous literature has deemed to be important for providing for trans* clients. It is reassuring that these programs are able to cover the majority of these topics in their education, as the literature on genetic counseling

for trans* individuals have cited pedigree nomenclature,^{44,51,53,56} disclosure of trans* status,^{48,49,52} appropriate use of language,^{20,42,51,47} risk assessment,^{45,47,57,58,60} and unique psychosocial factors^{40,51,60-62} as vital components of counseling trans* individuals effectively in a genetic counseling session.

Respondents who do not currently incorporate education on trans* healthcare cited a lack of appropriate educational materials as a barrier to offering this education in their programs. Therefore, access to these materials is a limiting factor for programs in the decision to offer education on trans* genetic counseling. Access to this material, therefore, may be influential in allowing these programs to offer this education in the future. Of note, lack of access to educational material is not the only barrier to offering this education, as two out of four programs cited lack of knowledgeable faculty/staff and one out of four cited lack of available time in the program as additional barriers. One program noted that it had not previously occurred to them to consider incorporating this education prior to the past few years, when it had become apparent that there may be unique considerations for this population. Therefore, this project may help to minimize one of several barriers programs are facing when thinking about incorporating trans* genetic counseling issues into their curriculum. No programs felt that education on trans* genetic counseling was unnecessary/irrelevant to genetic counseling training, and no respondents cited student disinterest as a reason for not offering this material. Additionally, 95% (19/20) of respondents either agreed or strongly agreed with the statement that “Genetic counseling programs should incorporate education on counseling transgender/gender nonconforming individuals into their curriculum.” This may suggest that while programs may not uniformly agree that it should be a requirement enforced by ACGC, most feel that they should be teaching their students on trans* genetic counseling issues. This, with the fact that most programs already offer some

education in this area, suggests that the majority of genetic counseling programs understand the importance of trans* genetic counseling competencies, whether or not they are currently able to incorporate these topics into their educational programs.

3.4.2 Perceived benefit of educational resource materials

Though the majority of responding programs already implement education for counseling trans* clients, 85% of programs desire access to additional educational resource materials on the topic. This result from genetic counseling program leadership is consistent with the fact that many practicing genetic counselors have expressed desire for more education on genetic counseling for trans* individuals, within clinical settings or within their training programs.^{46,47,53,63} The genetic counseling workforce is asking for more education on genetic counseling for trans* individual and beginning this support in training programs will help meet this need. Additionally, this desire for educational materials on these topics and the recognition that this curriculum is important to learning aligns with the ACGC's designation of multicultural competency as a required component of every genetic counselors' psychosocial education, as well as a necessary competency for any practicing genetic counselor.⁶⁷

Though the majority of respondents said that access to more educational resource material would be helpful, 10% (2/20) responded that they were unsure whether this access would be helpful, and 5% (1/20) responded that they would not be helpful. The respondent who did not feel access to more materials would be helpful was prompted to explain the response and indicated that the program they are affiliated with has a "fairly robust" curriculum related to LGBT and trans* issues in genetic counseling, and therefore access to these materials would likely not significantly influence the education already in place for the program's students. No programs responded that

they would not benefit from access to these materials because they would be irrelevant to their programs' curriculum. Therefore, these findings suggest that all or nearly all responding programs have an understanding of the importance of cultural competency in this area.

Of those programs who responded that additional educational resource materials would be or may be helpful, the types of materials perceived to be the most helpful were clinical case examples (100%) and role-playing scenarios (94.7%), suggesting the importance of hands-on learning and learning by example in genetic counseling programs. More educational resource materials, such as peer-reviewed articles from research with trans* individuals (73.7%), resources for providers working with trans* clients (73.7%), and a list of resources that are helpful for trans* patients (73.7%) were also perceived to be helpful for genetic counseling programs. This information is aligned with genetic counseling educators who have emphasized learning through experience/example and use of educational materials to suit multiple learning styles.¹⁰ The resources least selected were PowerPoint slides and premade quizzes on trans* genetic counseling topics. Though these educational resource materials can be useful and are likely widely used in genetic counseling programs, the results of this survey may be indicative of a move away from more traditional didactic strategies like lecture-style teaching and an emphasis on practical knowledge and experiential learning, which have been emphasized within and vital to genetic counseling education since these programs were established.⁷³ While traditional learning methods are important to a developing genetic counselor, experiential work in clinicals and simulations offer the most opportunity for hands-on professional development.⁷⁴

For the purposes of this study, an additional counseling for trans* clients fact sheet was developed with information on counseling this population to guide learners since didactic materials were not commonly selected by study participants. This educational resource guide as well as the

additional educational materials were created using information on genetic counseling for trans* individuals elicited from primary literature, professional guidelines for other clinical healthcare professions, guidelines developed by trans* advocacy organizations, and data collected by the survey. Information on barriers to healthcare and perspectives from trans* clients on interacting with healthcare professionals in general was assessed based on the results from the National Transgender Discrimination Survey.⁵ General recommendations for trans* healthcare were obtained from resources such as The Joint Commission, a professional organization which exists to create standards and accreditations in healthcare in order to inspire all providers to continually improve their knowledge and quality of care,²⁵ as well as from professional organizations such as the American Counseling Association, which have their own sets of published guidelines for working with trans* clients in their fields.²⁸ Recommendations for eliciting information on gender and sex from clients were informed by guidelines for clinical health professionals^{48,49} with genetic counseling-specific considerations from the primary literature.⁴⁴ Information for pedigree nomenclature came from guidelines published by the National Society of Genetic Counselors⁵⁵ and the National Comprehensive Cancer Network⁵⁴ as well as primary literature in genetic counseling with trans* individuals.⁴⁴ Strategies for competent use of language during a session were informed by primary literature within genetic counseling^{44,51} as well as by literature and published guidelines for professional organizations such as the American Counseling Association.^{5,28} Likewise, psychosocial considerations included in the educational resource material were curated from genetic counseling primary literature⁵¹ and professional guidelines.^{40,62} Cancer recommendations and considerations were mainly informed by published genetic counseling case examples and primary literature which assess genetic counselors' comfort with counseling trans* patients in a cancer setting^{45,47,58} as well as by primary literature in the field of cancer management

and care.⁵⁹⁻⁶¹ The full review of literature was used to develop these materials, with focus on the most recent and agreed upon recommendations and strategies.

3.4.3 Limitations

Several limitations to this study exist. First, voluntary response bias makes it more likely that those programs which are already invested in genetic counseling for trans* individuals are the ones who would take the time to respond to this survey, meaning that the sample of programs reached by this study may be more likely to have education on and experience with trans* genetic counseling than the total population of genetic counseling programs. Additionally, the 20 responses received by this survey represent approximately 35% of all ACGC accredited genetic counseling programs. Lastly, as discussed, older, more established programs in major cities appear to be overrepresented in the sample of programs. These limitations may diminish the generalizability of the study findings.

The educational resource materials created for this project are not all-encompassing and may not cover every consideration unique or important to genetic counseling for trans* individuals. Additionally, as the trans* community grows and more research is done within this population, the recommendations made and skills exercised through these materials may become outdated or inaccurate. These materials may not be as suitable for individuals who benefit from didactic learning tools such as Powerpoint presentations and formal evaluations.

3.4.4 Future Directions

For the scope of this thesis project, only the six educational resource materials perceived to be the most helpful by respondents were created for programs to access. However, even those materials could enhance a well-rounded education regarding trans* health issues and were thought to be helpful by several of the surveyed programs. . Future projects may use the results of this study to create other materials perceived to be beneficial to the education of genetic counseling students. Additionally, more involved in-person projects and seminars would likely be helpful and can be synthesized around these materials.

Additionally, it would be beneficial to evaluate the educational resource materials should genetic counseling programs choose to implement them into their program curriculum. While the scope of this project is to create educational resource materials, ultimately the goal of this study is to make genetic counselors more effective and culturally competent counselors for gender minorities. Consequently, a future project could focus on assessing whether or not these materials were helpful for students, and whether they learned from the available resources. A survey could be given to programs who incorporated these materials into their program education to elicit their perspectives on the effectiveness of the materials in educating students, or some type of student assessment could be administered to evaluate student learning. This feedback could be used to improve and expand upon the materials made as a part of this thesis project, as well as to assess the efficacy of the materials. Ongoing research within and conversations with trans* populations will help improve these materials with more perspectives from the target patient population in the literature. These materials should be considered an ongoing project with room for updates as terminology and perspectives change on trans* issues. Additionally, feedback should be elicited

from trans* individuals on these educational resources, and individual perspectives could be incorporated into the materials in order to serve a broader population of trans* individuals.

The scope of this study covers increasing access to educational materials for use during the genetic counseling master's degree program. Moving forward, it would be beneficial to think about educational opportunities and materials that would benefit practicing genetic counselors in counseling trans* clients as well.

3.5 Conclusion

This study is the first to assess education on genetic counseling for trans* populations currently employed by ACGC-accredited genetic counseling programs. Survey results demonstrate that most genetic counseling programs provide education on trans* issues in genetic counseling through course material, internship experiences, and other educational methods such as roleplay opportunities and review of case vignettes. The overwhelming majority of respondents felt that genetic counseling programs should incorporate education on counseling trans* individuals into their curriculum, regardless of their current programming. The majority of programs also indicated that access to additional educational resource materials on genetic counseling for trans* individuals would be beneficial and some programs reported this lack of access as a barrier to providing education to students. This study aimed to create educational materials genetic counseling programs found helpful in an effort to increase access to education on trans*-specific considerations in genetic counseling with the ultimate goal of improving genetic counselors' care of trans* populations. Programs will have convenient access via the shared

AGCPD resource drive to the materials made for this study and may choose to integrate them into their curriculum.

4.0 Research Significance to Genetic Counseling and Public Health

Lesbian, Gay, Bisexual, and Transgender (LGBT) individuals often experience disparities and discrimination in healthcare due to their sexual and gender minority status. Trans* individuals may experience different barriers from sexual minorities in part due to the fact that they have more interactions with and dependence upon the healthcare system for transition-related purposes,²¹ and in part because transphobia is pervasive.⁵ Multiple organizations such as the American Public Health Association and the 2020 Healthy People campaign have recognized the need to address the health disparities trans* people in America face.^{3,4} LGBT (Lesbian, Gay, Bisexual, and Transgender) populations experience higher rates of substance use, tobacco use, and obesity than the population average,⁴ and mental health concerns such as depression and anxiety appear to be elevated specifically in the trans* population,⁵ as 41% of trans* respondents to a national survey reported that they had tried to complete suicide, four times the national average.⁵ Trans* individuals are additionally more likely than cisgender individuals to experience interpersonal violence and are much more likely to delay important medical treatment out of fear of discrimination.⁶ In addition, trans* people are more likely to be uninsured, and experience unemployment, homelessness, legal discrimination in adoption and marriage, and loss of support from parents or family members, all of which are considered to be social determinants of health.⁵

Genetic counseling is defined by the National Society of Genetic Counselors (NSGC) as “the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.”⁸ While many efforts have been made to promote and teach multicultural competency in genetic counseling, like all other forms of healthcare in the United States, inequities in access to genetic counseling and quality of care persist for minority

populations.⁹ One of the three core functions of public health is assurance, which includes the assurance of a competent workforce capable of delivering high quality care to all people.⁷⁵ Genetic counseling has the potential to benefit trans* individuals seeking guidance on their risk for genetic conditions, reproductive risks and options, and medical decision making. Genetic counseling is a medical service in which psychosocial care of the patient is emphasized and client rapport is vital. Genetic counselors often function to provide resources to clients facing many of the psychological, social, and financial disparities many trans* individuals do at disproportionate rates.¹⁰

Genetic counselors and professional health organizations have already begun the work of identifying trans* health specific counseling strategies and resources associated with more positive healthcare outcomes through research and clinical experiences. The literature on genetic counseling for trans* individuals has cited pedigree nomenclature,^{44,51,53,56} disclosure of trans* status,^{48,49,52} appropriate use of language,^{20,42,51,47} risk assessment,^{45,47,57,58,60} and unique psychosocial factors^{40,51,60-62} as some facets integral to the process of genetic counseling that there are unique considerations for when it comes to providing competent and quality care to trans* clients. Each of these counseling issues may be complicated by or approached in a unique way for clients who are trans*. A recent study of genetic counselors found that around half felt less than confident about their ability to lead a session with a trans* client.⁴⁷ Additionally, multiple studies have found an overwhelming desire from genetic counselors for more education, both during their time in school and post-graduation, on counseling trans* and LGBT individuals.^{46,47,53,63} Promoting knowledge on these complexities in genetic counseling programs can help to ensure that newer generations of counselors are informed on appropriate counseling strategies and are therefore more well equipped to provide for their trans* clients.

Healthy People 2020 cited a lack of knowledgeable providers for LGBT individuals as one of the most prominent barriers negatively affecting the health of trans* people.⁴ One of the three core functions of public health is assurance, which includes the Essential Public Health Service of assurance of a competent workforce capable of delivering high quality care to all people.⁷⁵ This study adds to the genetic counseling literature as the first to examine trans* genetic counseling related content utilized in ACGC-accredited genetic counseling programs. Additionally, the study contributes access to educational resource materials via AGCPD to these programs which can potentially be incorporated into the training of genetic counseling students, ideally building a stronger workforce of individuals trained to work with trans* clients in mind. Education on trans* health in professional school settings for clinical health professionals has been shown to positively influence providers' confidence in their ability to provide culturally competent service and to improve outcomes for trans* patients.⁶⁴⁻⁶⁶ Including this type of education creates more informed future genetic counselors, which addresses the current workforce's desire for more opportunities to explore this topic. Another of the Essential Public Health Services is to assure an effective system that enables equitable access to the individual services and care needed to be healthy.⁷⁵ Ultimately, the goal of this study within the field of trans* genetic counseling is to impact public health by decreasing the health disparities trans* people face due to discrimination or ignorance of healthcare providers, thereby creating access to more equitable and competent care from providers trained to counsel diverse populations. By increasing genetic counselors' knowledge of genetic counseling for trans* clients, their ability to provide quality, competent care to this client base will hopefully improve.

Appendix A Invitation to Participate in Survey

Dear genetic counseling program leadership,

My name is Kaylee Williams and I am a second year genetic counseling student at the University of Pittsburgh. I'm reaching out to you all today to invite you to participate in a research study.

I am conducting an online, anonymous survey as part of my master's thesis project entitled "Genetic Counseling for Trans* Populations: An Educational Tool for Genetic Counseling Programs." The survey should take about **10 minutes** to complete and only **one response per program is needed**. This research project seeks to elicit currently implemented curricular content regarding genetic counseling for transgender and gender non-conforming individuals in ACGC accredited programs. The overall aim of this project is to create educational resource material that can be distributed to and used by genetic counseling programs.

To participate in the survey, please use this link: (link to survey provided here).

This project is being conducted by Kaylee Williams in partial fulfillment of a master's degree in Genetic Counseling at the University of Pittsburgh. This study has been approved by the University of Pittsburgh Institutional Review Board, STUDY20080052.

Thank you for your time and participation. Should you have any comments or questions, please feel free to contact me at knw30@pitt.edu.

Appendix B Survey

Introduction

This survey has been created as part of a masters research project entitled “Genetic Counseling for Trans* Populations: An Educational Tool for Genetic Counseling Programs.” This research project seeks to elicit currently implemented curricular content regarding genetic counseling for transgender and gender non-conforming individuals in ACGC accredited programs. The overall aim of this project is to create educational resource material for use by genetic counseling programs.

Only one response per program is needed. Please answer the following questions regarding your program’s current practices regarding education for counseling transgender and gender nonconforming clients, as well as what education materials may be valuable to you. This survey should take about **10 minutes** to complete. There are no foreseeable risks or benefits associated with participation in this project. You will not receive compensation for participation. All answers will remain voluntary, anonymous, and confidential. Results will be stored in a private manner. Your participation is voluntary, and you may choose not to finish the survey at any time. Should you have any comments or questions, you can contact Kaylee Williams at knw30@pitt.edu.

The following paragraph provides background information and relevant terminology which may be helpful to review prior to taking the survey.

Individuals who identify as **transgender** do not identify with the sex they were assigned at birth. A **transgender woman** is an individual who was assigned male at birth, but currently identifies as a woman. A **transgender man** is an individual who was assigned female at birth, but currently identifies as a man. In contrast, those who are **cisgender** identify with the sex they were assigned at birth. The term **gender nonconforming** encompasses individuals who do not express their gender outwardly in a way that conforms to what people might expect based on the sex they were assigned at birth. Transgender identity and gender nonconforming identity are not mutually exclusive. Importantly, the questions in this survey are not meant to include individuals born with **disorders of sex development**, a group of medical conditions characterized by problems with sexual development and sex differentiation which have genetic causes.

Block 1: Accreditation

Q1 Is your affiliated genetic counseling program ACGC-accredited?

- a) Yes
- b) No

Condition: No Is Selected. Skip To: End of Survey

Block 2: Program Education

Q2 Does your genetic counseling program provide education to all students on counseling for LGB (Lesbian, Gay, Bisexual) individuals? Please consider only educational content on sexual minorities which is not directly related to counseling for transgender and gender nonconforming individuals.

- a) Yes
- b) No

Q3 Does your genetic counseling program provide education to all students on counseling for transgender and gender nonconforming individuals? Please do not include any education done for counseling individuals with Disorders of Sex Development in your considerations.

- a) Yes
- b) No

Condition: No Is Selected. Skip to: Q9

Q4 The following questions ask you to consider your program's approach to gender identity as part of the curriculum. This should not include your program's approach to sexual orientation. Why does your program provide education for counseling transgender/gender nonconforming individuals? Please check all that apply.

- a) The genetic counseling program faculty feels it is important to include as part of students' education
- b) Transgender health education is required by the institutional body the program is a part of
- c) Other: please explain

Q5 What educational methods are used to teach students about counseling for transgender/gender nonconforming individuals? Please check all that apply.

- a) One lecture on counseling transgender/gender nonconforming individuals
- b) Greater than one lecture on counseling transgender/gender nonconforming individuals
- c) Content on counseling transgender/gender nonconforming individuals within another lecture or multiple other lectures
- d) Assigned case reports for counseling transgender/gender nonconforming individuals
- e) Simulated role plays for counseling transgender/gender nonconforming individuals
- f) Required reading on counseling transgender/gender nonconforming individuals
- g) Optional reading on counseling transgender/gender nonconforming individuals
- h) Required web-based tutorials for counseling transgender/gender nonconforming individuals
- i) Optional web-based tutorials for counseling transgender/gender nonconforming individuals
- j) Required documentary, film, or video about counseling transgender/gender nonconforming individuals
- k) Optional documentary, film, or video about counseling transgender/gender nonconforming individuals
- l) Other: please describe

Q6 Who primarily delivers this educational material for genetic counseling for transgender/gender nonconforming individuals? Please choose all that apply.

- a) Healthcare provider other than a genetic counselor with experience in caring for transgender/gender nonconforming individuals
- b) Genetic counselor
- c) Individual from a transgender health advocacy group
- d) Other instructor: please explain

Q7 Is this educational material for counseling transgender/gender nonconforming individuals presented in the first or second year of a program?

- a) First
- b) Second
- c) Material is presented across both years

Q8 Which topics on transgender health are generally covered by this educational material? Please select all answers that apply and add any topics that are covered which are not listed here.

- a) Health disparities faced by transgender and gender nonconforming individuals
- b) Barriers to care faced by transgender and gender nonconforming individuals
- c) Transgender identities
- d) The difference between sex and gender identity
- e) Use of appropriate pronouns
- f) Use of gender-neutral language
- g) Eliciting gender identity, appropriate pronouns, and sex assigned at birth from transgender/gender nonconforming patients
- h) Possible pedigree nomenclature for transgender/gender nonconforming individuals
- i) Risk assessment for transgender/gender nonconforming individuals
- j) Psychosocial considerations for transgender/gender nonconforming individuals
- k) Personal bias training
- l) Other:

Condition: Question is Answered. Skip To: End of Block.

Q9 Why does your program not provide education on genetic counseling for transgender/gender nonconforming individuals? Please check all that apply.

- a) Lack of knowledgeable faculty/staff
- b) Lack of available time in the program to cover this material
- c) Lack of appropriate educational materials
- d) Students are disinterested
- e) The education is unnecessary/irrelevant to genetic counseling training
- f) Other reason: please describe

Q10 Please choose the one that is the most important factor in why your program does not provide education on genetic counseling for transgender and gender nonconforming individuals:

- a) Lack of knowledgeable faculty/staff
- b) Lack of available time in the program to cover this material
- c) Lack of appropriate educational materials
- d) Students are disinterested
- e) This education is unnecessary/irrelevant to genetic counseling training
- f) Other reason specified in previous question

Block 3: Educational Utility/Need

Q11 Would your program benefit from access to educational resource material concerning genetic counseling for transgender/gender nonconforming patients?

- a) Yes
- b) No
- c) Unsure/Maybe

Condition: Yes or Unsure/Maybe Is Selected. Skip To: Q13

Q12 If no, please briefly explain why you feel your program would not benefit from access to these educational materials.

Condition: If this question is Not Empty. Skip to: Q14

Q13 What types of educational material regarding counseling for transgender/gender nonconforming populations would be most helpful? Please select as many as you believe would be beneficial to your program.

- a) Powerpoint slides with speaker notes
- b) Clinical case examples that highlight transgender/gender nonconforming-specific considerations in genetic counseling
- c) A pamphlet/guidebook with general information about counseling transgender/gender nonconforming populations
- d) List of peer-reviewed articles on transgender and gender nonconforming individuals' recommendations to genetic counselors
- e) List of peer-reviewed articles on genetic counselors' recommendations on working with transgender and gender nonconforming individuals
- f) Role playing scenarios that can be used to practice learned skills
- g) Quizzes on relevant topics in providing competent care for gender minorities for genetic counselors
- h) List of websites and organizations that provide valuable resources for working with transgender/gender nonconforming clients
- i) List of available patient resources for transgender and gender nonconforming clients
- j) Any other recommendations, please describe:

Q14 Currently, the Accreditation Council for Genetic Counseling (ACGC) Standards for Accreditation lists "multicultural sensitivity and competency" as a required component of genetic counseling education (ACGC Standards of Accreditation for Graduate Programs in Genetic Counseling 2019). The ACGC Practice-Based Competencies for Genetic Counselors lists the ability to "Apply genetic counseling skills in a culturally responsive and respectful manner to all clients" as a skill that competent genetic counselors should master (ACGC Practice-Based Competencies for Genetic Counselors 2019). Please indicate the degree to which you agree or disagree with the following statement: the concept of multicultural competency includes gender minorities such as transgender and gender nonconforming populations.

- a) Strongly agree
- b) Agree
- c) Neither agree nor disagree
- d) Disagree

- e) Strongly disagree

Q15 Currently, the Accreditation Council for Genetic Counseling (ACGC) Standards for Accreditation lists “multicultural sensitivity and competency” as a required component of genetic counseling education (ACGC Standards of Accreditation for Graduate Programs in Genetic Counseling 2019). The ACGC Practice-Based Competencies for Genetic Counselors lists the ability to “Apply genetic counseling skills in a culturally responsive and respectful manner to all clients” as a skill that competent genetic counselors should master (ACGC Practice-Based Competencies for Genetic Counselors 2019). No specific mention is made of counseling for gender minorities. Should ACGC require that all genetic counseling students acquire the ability to counsel gender diverse populations?

- a) Definitely yes
- b) Probably yes
- c) Unsure
- d) Probably not
- e) Definitely not

Q16 Please indicate the degree to which you agree or disagree with the following statement: Genetic counseling programs should incorporate education on counseling transgender/gender nonconforming individuals into their curriculum.

- a) Strongly agree
- b) Agree
- c) Neither Agree nor Disagree
- d) Disagree
- e) Strongly disagree

Block 4: Program Demographics

Q17 How long has your genetic counseling program been in existence?

- a) 0-3 years
- b) 4-9 years
- c) >10 years

Q18 In which region of North America is your program located?

- a) Region 1 (CT, MA, ME, NH, RI, VT, CN Maritime Provinces)
- b) Region 2 (DC, DE, MD, NJ, NY, PA, VA, WV, PR, VI, Quebec)
- c) Region 3 (AL, FL, GA, KY, LA, MS, NC, SC, TN)
- d) Region 4 (AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI, Ontario)
- e) Region 5 (AZ, CO, MT, NM, TX, UT WY, Alberta, Manitoba, Saskatchewan)
- f) Region 6 (AK, CA, HI, ID NV, OR, WA, British Columbia)

Q19 In general, how many students are accepted each year to your institution’s genetic counseling program? Some schools have decided to accept fewer applicants for the 2020-2021 school year than they normally would have due to the COVID-19 pandemic. Please do not take these circumstances into consideration for this question, and instead answer with the class size which your program can normally accommodate.

- a) 2-4 students

- b) 5-10 students
- c) 11-18 students
- d) 18+ students

Q20 Please select the option that best describes the environment in which the program is situated.

- a) Major city (Population of at least 800,000)
- b) Large city (Population 500,000 to 800,000)
- c) Medium city (Population 100,000 to 500,000)
- d) Small city (Population 50,000 to 100,000)
- e) Town/Rural Environment (Population 50,000 or less)

Q21 Which choice most accurately reflects the format of the classes offered by your program? Please do not include online accommodations made due to closures in the past year from the COVID-19 pandemic.

- a) No part of the curriculum is presented online
- b) Less than 50% of the curriculum is presented online
- c) Curriculum is evenly presented online and in person
- d) Between 51-75% of the curriculum is presented online
- e) Nearly 100% of the curriculum is presented online

End Survey

Appendix C Genetic Counseling for Trans* Individuals Educational Resource Materials

Appendix C.1 Genetic Counseling for Transgender Individuals Educational Resource Guide

Transgender Identities and Terminology

Term	Definition
Sex assigned at birth (SAAB) (n.):	A designation of male or female (as well as intersex in some states) assigned to babies at birth usually based on external genitalia
Gender identity (n.):	An internal and personal understanding of one's own gender which may or may not correspond with biological sex
Transgender (adj.):	A descriptor for a person whose gender identity is incongruent with or different from the sex they were assigned at birth <ul style="list-style-type: none">Previously used terms such as transsexual are no longer commonly used or appropriate
Cisgender (adj.):	A descriptor for a person whose gender identity is congruent with or the same as their sex assigned at birth
Nonbinary (adj.):	An umbrella term for individuals whose gender identity is not described by the binary identities of man and woman
Gender nonconforming (GNC) (adj.):	A descriptor for individuals whose behavior, physical traits, cultural norms, and/or presentation of gender is socially perceived to be incongruent with the sex they were assigned at birth
Gender neutral (adj.):	A descriptor for a word or expression that can be applicable to all genders—usually implies that no one gender is specified
Gender dysphoria (n.):	A psychologically painful phenomenon produced by the discord between one's gender identity and sex assigned at birth <ul style="list-style-type: none">Not all transgender individuals experience gender dysphoria
AMAB (adj.):	An acronym for assigned male at birth—refers to a person whose assigned biological sex is male
AFAB (adj.):	An acronym for assigned female at birth—refers to a person whose assigned biological sex is female

General Counseling Strategies

Disclosure of trans* status

- Eliciting sex assigned at birth and gender identity in a competent way is beneficial to maintaining rapport²⁵ and is vital to proper risk assessment in some cases
- Strategies that can help clients feel comfortable disclosing gender identity and SAAB:
 - An LGBT-friendly clinic environment⁴⁸

- Symbols specific to transgender inclusion are more highly appreciated⁴⁴
- An explanation of why this information is important⁴⁸
- An understanding that this information is routinely collected from every patient^{48,49}
- Good rapport building means a higher likelihood that clients will trust you with this information

Pedigree nomenclature

- There is currently no agreed upon standardized pedigree nomenclature for transgender individuals
 - The National Comprehensive Cancer Network (NCCN) recommends using a circle inside a square for trans men, and a square inside a circle for trans women, with no recommendations for nonbinary clients⁵⁴
 - The National Society of Genetic Counselors (NSGC) recommends using the symbol that corresponds to the patients' gender with notation of their (presumed) karyotype (XY or XX)⁵⁵
 - A recent study found that transgender individuals prefer the use of a symbol that corresponds to their gender identify (using a diamond for nonbinary genders) with notation of SAAB (AFAB or AMAB) below the symbol⁴⁴
 - Remember that often a client can see you drawing the pedigree, and in some cases, it is helpful to show the pedigree to the client
 - It is important that the client feels represented by the symbol used on the pedigree for psychosocial care and rapport—using a gender-affirming symbol can enhance the rapport building process. You can consider asking the client what symbol feels the most accurate to them.

Appropriate use of language

- It's important to use a patient's proper pronouns and name when counseling^{28,51}
 - In the event that the wrong name or pronoun is used, apologize briefly and make it a point to not misgender the client again
- Gender-neutral language can be employed in all counseling sessions but is particularly relevant for counseling transgender clients, who tend to prefer specific, scientific language that is not gendered^{24,44}
 - Refer specifically to anatomical structures, such as “people with uteruses” rather than “women” or “females”²⁴
 - Using chromosomal makeup rather than sex to describe sex modified risks—for instance, saying “you are at an increased risk because you have an X and Y chromosome” rather than “you are at an increased risk because you are male”⁵¹
- Take time to learn about and become comfortable speaking about the difference between gender and sex^{44,51}
 - Many transgender individuals express frustration or exhaustion over having to explain their gender identity to providers⁵

- It's okay to ask clients questions when something is unclear but try to familiarize yourself with basic terminology surrounding transgender identities

Unique psychosocial factors

- Being transgender is not a mental illness.
- Use psychosocial counseling tools and non-directive counseling as you would for any patient—allow individuals to speak from their experience, and validate/take into account those experiences during the session⁴⁰
- Transgender individuals report higher rates of anxiety and depression than cisgender individuals⁴⁰
- Transgender individuals are more likely to experience a lack of familial support and may have non-traditional familial structures or “chosen” families, which may make topics such as family history of disease a sensitive topic⁵¹
- Assess transgender clients’ support network where appropriate
 - Increased social support has been linked to better coping skills, lower risks of anxiety and depression, and overall better healthcare outcomes for transgender patients⁶²
- Using proper pronouns, names, and terminology with a patient results in better psychosocial outcomes⁵¹

Specialty-specific Approaches and Considerations

Cancer

- Counseling for cancer predisposing syndromes is complicated by differing motivations for testing, past gender-affirming surgeries, and the decision to pursue hormone replacement therapy (HRT).^{45,47}
- Transgender individuals may have unique reasons for desiring cancer genetic testing.⁴⁷
 - Transgender individuals may pursue testing at younger ages in an attempt to obtain coverage for “top surgery” in the form of having a prophylactic mastectomy⁵⁸
 - Be aware that typical top surgery or gender affirming male chest reconstruction preserves more breast tissue than a prophylactic mastectomy
 - Transgender individuals may pursue testing in order to have more information on risks of estrogen and progesterone HRT on their risk for cancers⁵⁸
 - Current data on the risk for cancers in individuals using estrogen hormone replacement therapy is inconclusive—some studies report breast cancer risks for trans* individuals taking estrogen are heightened over the general population of AMAB individuals, and some report no significant elevation of risk⁵⁹⁻⁶¹
- Take into account the physical and mental wellbeing of a transgender patient when discussing cancer care and transition care options⁵⁰

- Example: transgender individual using gender-affirming estrogen is diagnosed with breast cancer—a discussion about stopping estrogen therapy is relevant, but providers should keep in mind the health benefits of gender affirmation and the mental health of the patient as well^{60,61}

Pediatrics

- Many times, transgender youth do not share their gender identities with their families for their own safety, or because they are not ready to
- Parents of transgender individuals may not use the correct names/pronouns for transgender children—counselors can choose to explore this with families where appropriate
- Think about use of gender neutral and competent language, especially when discussing X-linked conditions and pediatric conditions modified by sex

Prenatal

- Pregnancy is a highly gendered experience in the United States
 - The imagery associated with and terminology used when talking about pregnancy is usually meant for cis women
 - “Women’s clinics” may be a difficult or uncomfortable place for transgender individuals to visit
- Some transgender individuals may have to halt or delay medical transitional care in order to have children.
 - This can be a difficult experience and can enhance feelings of gender dysphoria for some people
- For preconception appointments, take time to familiarize yourself with current options for reproduction for transgender individuals³⁸

Appendix C.2 Genetic Counseling for Trans* Individuals Clinical Case Studies/Roleplays

Vignette #1: Pediatric Genetic Counseling Case

Victor Young is a 16-year-old male referred to medical genetics for suspicion of a connective tissue disorder. Victor’s mother accompanies Victor to the appointment. As the genetic counselor facilitates introductions, Victor informs the counselor that she prefers to go by Viola. The counselor thanks Viola for sharing her preferred name and asks Viola whether there are pronouns she uses, to which Viola responds she/her/hers. The counselor shares her pronouns as

well. While this exchange is happening, the counselor notices that the mother's affect has changed—she appears upset, has crossed her arms, and rolled her eyes when Viola shared her preferred name. The counselor begins the session.

Once, while going over Viola's medical history, the counselor calls Viola by the name she was given at birth. The counselor corrects herself, quickly apologizes, and moves on with the session without making the same mistake again.

During the session, Viola's mother continues to use he/him/his pronouns and Victor when referring to Viola. The counselor pauses the session and says, "Mrs. Young, I've noticed that you use a different name for Viola than she uses for herself." Mrs. Young replies, "Yes, it's been hard to adjust to calling him Viola. I still see him as my son." The counselor responds, "Change can be difficult to adjust to, especially when it comes to a person's children. Viola, what are your thoughts?" Viola pauses, and says, "I know that this change has been hard for my parents, but it hurts when they use the wrong name for me. I see myself as Viola, and I wish my mom did too." Mrs. Young's expression visibly softens. The counselor spends a bit longer exploring these emotions, and says, "Sometimes, it can be the job of a genetic counselor to start tough conversations like this one. This sounds like a conversation that will continue beyond this appointment, and I want to thank you both for exploring it with me for a bit today. How can I best support you as you continue to have these conversations?" The three discuss for a while longer, during which time the family decides they would benefit from information about family counseling from a trans*-friendly provider. The genetic counselor offers to do some research and give the family resources for this moving forward.

The counselor thanks Viola and her mother for their time at the end of the session and informs the geneticist of Viola's preferred name and pronouns prior to her consultation with the

doctor. The counselor also makes note of Viola's preferred name and pronouns in the medical record.

Discussion questions:

- What counseling strategies did the counselor use to affirm Viola's gender and build rapport? How did the counselor validate the mother's feelings while advocating for Viola?
- The counselor misgendered Viola once during the appointment—what did the counselor do to ensure that she maintained her rapport with the client? Would you have done anything differently?
- What are the possibilities and limitations that counselors face when advocating for a pediatric trans* patient?
- **Roleplay opportunity:** In groups of 3, roleplay the conversation about Mrs. Young's use of Viola's incorrect name and pronouns as Viola, the counselor, and Mrs. Young. Take turns being the counselor in this scenario, starting with the counselor's direct question to Mrs. Young about why she uses different pronouns for Viola. Discuss with your classmates what went well, and what additional counseling strategies could be used in this situation.

Vignette #2: Cancer Genetic Counseling Case

Alessia is an 18-year-old female who presents to cancer genetics for genetic testing for a known familial variant in BRCA2. Alessia's doctor has made a note in the referral that Alessia identifies as nonbinary and is pursuing this testing at a younger age than most patients in order to consider options about gender-affirming surgeries.

During the family history intake, Alessia becomes emotional and says they do not have any contact with their biological family. The counselor assures Alessia that though the family history

is an important tool for genetic counseling, it is okay if Alessia does not have all of the necessary information. During the family history, Alessia becomes visibly emotional and has trouble continuing to answer questions. The counselor notes Alessia's reaction to the family history and finishes up by asking only the most clinically relevant questions. The counselor validates Alessia's emotions when talking about their family and assesses Alessia's support system. Alessia makes it clear that though being estranged from their biological family has been difficult, they have a robust support system of transgender peers who have become their chosen family. Alessia's partner knows that they have a family history of cancer and has provided them support surrounding their motivations and anxieties about genetic testing.

Alessia discloses to the counselor that they secretly hope they are positive for the variant, as they do not believe they can afford gender-affirming "top" surgery without the insurance coverage for a prophylactic mastectomy. The counselor chooses to explore this statement further, and validates Alessia's motivations, saying, "Different people have different motivations for pursuing genetic testing, and it sounds to me like this testing will factor into the decisions you make or the options you'll have in the future for surgery." Alessia confirms this, and the counselor prompts, "How would you feel if the testing comes back negative?" Alessia replies, "I know it's messed up to be sad about it because I don't want to have cancer, but I would be more heartbroken if I found out I could not afford top surgery." The counselor explores these emotions with the client further, validating Alessia's motivations for testing and acknowledging that it would be difficult to learn that gender-affirming surgery is not an option. The counselor assures Alessia that she will provide support to them no matter what the outcome of the testing is and lets Alessia know that there are other ways of affording top surgery and community funds to assist them. Alessia seems visibly reassured by the counselor's words, and consents to the genetic testing. The counselor

thanks Alessia for their time and provides them with a list of resources on trans* health as well as some community funds for gender-affirming surgeries.

Discussion questions:

- How did the counselor adjust the counseling approach and customize the session to account for Alessia's family situation?
- What counseling interventions did the counselor use to ensure Alessia felt supported?
- **Roleplay opportunity:** In groups of 2, take turns imagining you are the counselor, and your partner, playing the role of Alessia, has just told you they secretly hope they are positive for the familial variant. How would you approach this conversation as the counselor?

Vignette #3: Prenatal Genetic Counseling Case

Emile is a 36-year-old FtM (Female to Male) individual presenting to prenatal genetics for advanced maternal age. A review of his chart reveals that Emile has previously been treated with testosterone. He arrives to the appointment with his partner, Cole. When the counselor retrieves Emile from the waiting room, it is clear that he and his partner are on edge. Cole introduces themselves, and when the counselor asks, Cole informs her that they are nonbinary and use they/them/theirs pronouns. The counselor makes small talk with the couple, and says, "I've noticed that you seem tense, is there anything on your mind that you'd like me to address before we begin?" Emile shakes his head no and says he is ready to move forward with the session. The counselor notes the couple's affect and moves on with the session.

During the entire session, the counselor is careful to use gender neutral and scientific terms over more gendered terms such as mother, father, male, and female. She uses "egg" and "sperm"

when discussing conception and refers to pregnant “individuals” rather than pregnant women. The couple visibly relaxes and asks questions throughout the appointment. When taking the family history, the counselor asks Emile whether Cole is a biological parent of the pregnancy, to which Emile responds yes. The counselor uses a square to represent Emile and writes “AFAB” under his symbol, and a diamond to represent Cole with “AMAB” under their symbol. The counselor goes through the family history, replacing phrases such as “how many brothers and sisters do you have?” with questions such as “do you have any siblings? Who are they?” to elicit information.

Once again, later in the session, the counselor asks the couple generally, “What are your concerns about this testing, or concerns that you have to share with me in general?” Emile replies, “This pregnancy has been very hard, and we’re just ready for our child to be healthy and here.” Emile expresses to the counselor that this pregnancy has been very difficult for the couple. Emile had to temporarily stop his use of exogenous hormone therapy in order to become pregnant and has found it is difficult to cope with the mood changes associated with halting his use of testosterone to be difficult to cope with. Emile expressed that the front desk staff at the office appeared confused and put off by his appearance. While in the waiting room, Emile and Cole felt watched by the pregnant women in the room and felt uncomfortable with the gendered pamphlets and informational booklets they were given by the front desk. Additionally, the ultrasound technician had misgendered Emile throughout their time together. The counselor acknowledges these concerns and validates that this time has been stressful for the pair. “It’s true that pregnancy is a very gendered process in this country, and I can see how this would be off-putting and frustrating for your family.” The counselor asks the couple if they have had any support during this time, and they respond that though friends and family have been supportive, they had been considering seeking out professional mental health care.

The counselor offers the couple some gender-neutral pregnancy resources as well as trans*-specific mental health services. The couple thanks the counselor, and after the session concludes, the counselor begins to ponder possible ways of improving trans* individuals' experiences with the clinic.

Discussion questions:

- What systemic changes can the counselor suggest to the rest of the clinic staff to make the clinic more trans*-inclusive?
- **Roleplay opportunity:** (Can be completed with 2 or 3 participants) Emile and his partner have just told you about what has been bothering them during this appointment. What counseling strategies would you use to establish ongoing rapport, validate the couple's frustrations, and provide support to the couple?

Appendix C.3 Patient Resources for Trans* Clients

UCSF Center of Excellence for Transgender Health (www.prevention.ucsf.edu/transhealth)

- Provides educational resources for healthcare workers and trans individuals with the goal of increasing access to comprehensive, effective, and affirming care for transgender communities

National Center for Transgender Equality (www.transequality.org)

- Provides advocacy, resources, aid, and resources for trans* individuals in the United States
- The Know Your Rights series outlines trans* individuals' legal rights in a healthcare setting

Out2Enroll (www.Out2Enroll.org)

- Describes resources to assist trans* individuals in selecting and enrolling in health insurance

Outcare (outcarehealth.org)

- Provides a nationwide resource for finding providers competent with working with LGBT individuals with opportunities for patients to leave reviews.

RAD Remedy (www.radremedy.org)

- Provides a community-sourced list of trans*-affirming healthcare providers

The Trevor Project (www.thetrevorproject.org)

- Supplies crisis intervention and mental health services for LGBT individuals between the ages of 13-24

TransChance Health (www.transchancehealth.org)

- Helps trans* individuals navigate healthcare and insurance and to receive high-quality affirming care

Transcend Legal (www.transcendlegal.org)

- Supplies a resource for helping trans* individuals get insurance coverage for gender-affirming medical expenses

TransHealth (www.trans-health.com)

- Provides guidance for healthcare providers and lists trans*-affirming health clinics

Appendix C.4 Provider Resources for Working with Trans* Clients

American Counseling Association Competencies for Counseling with Transgender Clients

- Describes competencies for use in psychological counseling with transgender clients with focus on wellness, resilience, and strength-based approach

American Psychological Association: Guidelines for Psychological Practice with Transgender and Gender Nonconforming People (<https://www.apa.org/practice/guidelines/transgender.pdf>)

- Provides information on the psychosocial care of trans* patients, originally constructed for long-term mental health specialists, but can be applied to a genetic counseling session

Association of American Medical Colleges' LGBT Health Resources (<https://www.aamc.org/what-we-do/diversity-inclusion/lgbt-health-resources>)

- Provides a variety of educational tools including videos meant to inform clinicians on appropriate standards of care for LGBT individuals

UCSF Center of Excellence for Transgender Health (www.prevention.ucsf.edu/transhealth)

- Provides educational resources for healthcare workers and trans individuals with the goal of increasing access to comprehensive, effective, and affirming care for transgender communities

Improved Patient Engagement for LGBT Populations: Addressing Factors Related to Sexual Orientation/Gender Identity for Effective Health Information Management – AHIMA Work Group

- Addresses considerations for improving practices and care provision for trans* patients

National Center for Transgender Equality (www.transequality.org)

Outcare (outcarehealth.org)

- Provides a nationwide resource for finding providers competent with working with LGBT individuals with opportunities for patients to leave reviews.

RAD Remedy (www.radremedy.org)

- Supplies a community-sourced list of trans-affirming healthcare providers

TransHealth (www.trans-health.com)

- Provides guidance for healthcare providers and lists trans-affirming health clinics

Appendix C.5 Peer-reviewed Articles on Genetic Counseling for Trans* Populations

Barnes H, Morris E, Austin J. Trans-inclusive genetic counseling services: Recommendations from members of the transgender and non-binary community. *Journal of Genetic Counseling*. 2019.

- Discussion with and recommendations from trans* individuals on appropriate pedigree nomenclature, trans-inclusivity, and disclosure of sex and gender identity in a genetic counseling setting.

Berro T, Zayhowski K, Field T, Channaoui N, Sotelo J. Genetic counselors' comfort and knowledge of cancer risk assessment for transgender patients. *Journal of Genetic Counseling*. 2019.

- Survey of cancer genetic counselors' education, knowledge, and comfort with trans* considerations and issues in cancer genetic counseling.

Glessner HD, VandenLangenberg E, Veach PM, LeRoy BS. Are Genetic Counselors and GLBT Patients "on the Same Page"? an Investigation of Attitudes, Practices, and Genetic Counseling Experiences. *Journal of Genetic Counseling*. 2012;21(2):326-336.

- Investigates perspectives and experiences from LGBT patients on genetic counseling as well as genetic counselors' attitudes and practices when counseling LGBT patients.

Lyninger HS. *Genetic Counseling for Transgender Patients: Perspectives on Terminology, Disclosure of Transgender Status, and Proposed Pedigree Nomenclature*. eScholarship, UC Irvine; 2019.

- Thesis project from a masters student which gathered perspectives from trans* individuals on their perception of genetic counseling, appropriate language and terminology, disclosure of trans* status, and pedigree nomenclature.

Sacca RE, Koeller DR, Rana HQ, Garber JE, Morganstern DE. Trans-counseling: A case series of transgender individuals at high risk for BRCA1 pathogenic variants. *Journal of Genetic Counseling*. 2019;28(3):708-716.

- Provides three case reports from genetic counselors' sessions with trans* individuals in a cancer genetics setting and extracts main themes and considerations for counselors.

Sheehan E, Bennett RL, Harris M, Chan-Smutko G. Assessing transgender and gender non-conforming pedigree nomenclature in current genetic counselors' practice: The case for geometric inclusivity. *Journal of Genetic Counseling*. 2019.

- Provides perspectives from genetic counselors on appropriate pedigree nomenclature for trans* individuals and emphasizes the need for standardized pedigree nomenclature for trans* patients.

Sutherland N, Espinel W, Grotzke M, Colonna S. Unanswered Questions: Hereditary breast and gynecological cancer risk assessment in transgender adolescents and young adults. *Journal of Genetic Counseling*. 2019.

- Identifies ways to appropriately assess cancer risk for trans* individuals as well as offers recommendations to counselors on psychosocial care and facilitating decision making.

VandenLangenberg E, Veach PM, LeRoy BS, Glessner HD. Gay, Lesbian, and Bisexual Patients' Recommendations for Genetic Counselors: A Qualitative Investigation. *Journal of Genetic Counseling*. 2012;21(5):741-747.

- Gathers information on patients' genetic counseling experiences and assesses their expectations for genetic counselors. While there are no trans* participants in this study, some findings from sexual minorities may be relevant to gender minorities.

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

- Examines interviews from a cohort of cancer genetic counselors who have worked with trans* patients in order to identify situations that may arise in clinic related to the patients' trans* status.

Appendix D IRB Approval



NOT HUMAN RESEARCH DETERMINATION

Date:	September 2, 2020
Review Type:	Initial Study
IRB:	STUDY20080052
PI:	Kaylee Williams
Title:	Genetic Counseling for Trans* Populations: Development of Educational Material for Genetic Counseling Programs

The Institutional Review Board determined that the proposed activity is not research involving human subjects as defined by DHHS and FDA regulations.

This determination applies only to the activities described in the IRB submission and does not apply should any changes be made. If changes are made and there are questions about whether these activities are research involving human in which the organization is engaged, please submit a new request to the IRB for a determination. You can create a modification by clicking **Create Modification / CR** within the study.

If you have any questions, please contact the University of Pittsburgh IRB Coordinator, [Carolyn Ivanusid](#).

Please take a moment to complete our [Satisfaction Survey](#) as we appreciate your feedback.

Appendix E Survey Data

Appendix Table A: Timing of Presentation of Trans* Genetic Counseling Materials

When Trans* Genetic Counseling Material is Presented in Genetic Counseling Programs	Respondents (n=16)
First year	2 (12.5%)
Second year	5 (31.3%)
Material is presented across both years	9 (56.3%)

Appendix Table B: Programs' Rationale for Implementing Trans* Counseling Education

Reason for Implementing Trans* Counseling Education	Respondents (n=16)
The genetic counseling program faculty feels it is important to include as part of students' education	16 (100%)
Transgender health education is required by the institutional body the program is a part of	0 (0%)
Other, please explain: Students have also made it clear that information to aid in providing an inclusive atmosphere for all individuals, particularly those who are trans/gender nonconforming is a priority.	1 (6.3%)
Other, please explain: Health disparities for transgender/gender non-conforming make this very important hence why we decided to include in our curriculum	1 (6.3%)

Appendix Table C: Instructors Utilized for Trans* Genetic Counseling Issues

Instructor(s) for Trans* Genetic Counseling Curriculum	Respondents (n=16)
Healthcare provider other than a genetic counselor with experience in caring for transgender/gender nonconforming individuals	6 (37.5%)
Genetic counselor	14 (87.5%)
Individual from a transgender health advocacy group	2 (12.5%)
Other instructor, please explain: MSSW social worker faculty member of GC program with clinical SW experience in working with transgender/gender nonconforming individuals	1 (6.3%)

Appendix Table D: Inclusion of Trans* Genetic Counseling in ACGC Accreditation Standards for Genetic Counseling Programs

Response: Should ACGC require that all genetic counseling students acquire the ability to counsel gender diverse populations?	Number of Programs N=20
Definitely yes	8 (40%)
Probably yes	2 (10%)
Unsure	6 (30%)
Probably not	4 (20%)
Definitely not	0 (0%)

Appendix Table E: Programs' Perception on Inclusion of Trans* Counseling Issues in Genetic Counseling Curriculum

Agree/Disagree: Genetic counseling programs should incorporate education on counseling transgender/gender nonconforming individuals into their curriculum.	Number of Programs N=20
Strongly agree	17 (85%)
Agree	2 (10%)
Neither agree nor disagree	1 (5%)
Disagree	0 (0%)
Strongly disagree	0 (0%)

Appendix Table F: Perceived Benefit of Access to Educational Resource Materials

Would your program benefit from access to additional educational resource materials on trans* counseling?	Respondents (n=20)
Yes	17 (85%)
Unsure/maybe	2 (10%)
No, please explain: We have a fairly robust curriculum in this area. We start in first year with a lecture, role play and journal club. In second year we have a guest lecturer genetic counselor who is does regular workshops on transgender and LGBTQ healthcare issues and genetic counseling.	1 (5%)

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