

**ORIGINAL ARTICLE**

# Assessing transgender and gender non-conforming pedigree nomenclature in current genetic counselors' practice: The case for geometric inclusivity

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**Abstract**

Healthcare professionals rely on national organizations for guidance; the National Society of Genetic Counselors (NSGC) and the National Comprehensive Cancer Network (NCCN) have differing guidelines for acceptable pedigree symbols to represent transgender patients and minimal recommendations for gender non-conforming (GNC) patients. Inconsistency in accepted pedigree symbols to represent these patients is a barrier to providing them appropriate care. We assess variability in pedigree practice among genetic counselors and students, as well as reported education on serving the needs of the transgender and GNC communities, through a survey distributed through NSGC. Participants felt symbols similar to NSGC's (41.1%) and NCCN's (29.7%) recommendations for transgender patients are appropriate and emphasized a desire to affirm gender identity. We identified greater variability in symbols representing a GNC patient; 19.2% of participants selected 'other', explaining they were unsure of the appropriate choice. A high interest (99%) in further training demonstrates a recognition of education as an effective strategy for improving awareness and competency. Promotion of existing resources could help address the fact that 81% of participants were unaware of any standardized symbols used to represent transgender individuals. Creating affirming, standardized pedigree nomenclature is necessary for appropriate and consistent care.

**KEYWORDS**

cultural competence, discrimination, disparities, diversity, education, gender non-conforming, genetic counseling, genetic counselors, genetics services, LGBTQ+, pedigree, psychosocial, stigma, transgender, underrepresented populations

**1 | INTRODUCTION**

The population of patients identifying as lesbian, gay, bisexual, transgender, and queer (LGBTQ+: the plus indicates additional gender and sexual minority populations not included in the original acronym) disclosing their gender identity and/or sexual orientation to their healthcare providers has increased over the last decade, prompting providers to seek out additional resources outside of their traditional training to provide appropriate care for these

patients (Fredriksen-Goldsen et al., 2014). The healthcare community is continuing to expand its knowledge of LGBTQ+ health; however, the experiences of transgender and gender non-conforming (GNC) individuals are often rendered invisible under the broad LGBTQ+ umbrella. The transgender and GNC populations represent a complex and diverse set of identities, and terminology used within the community can differ and evolve. For this study, we referred to the definitions offered by Fenway Health (Glossary of Gender & Transgender Terms, 2010; Table 1). Much of the existing literature

**TABLE 1** Glossary of terms pertaining to sex and gender identity as defined by Fenway Health (2010)

Term	Definition
Gender identity	'A person's innate, deeply-felt psychological identification as a man, woman, or something else, which may or may not correspond to the person's external body or assigned sex at birth'
Sex	'In a dichotomous scheme, the designation of a person at birth as either "male" or "female" based on their anatomy (genitalia and/or reproductive organs) and/or biology (chromosomes and/or hormones)'
Gender expression	'The external manifestation of a person's gender identity, which may or may not conform to the socially-defined behaviors and external characteristics that are commonly referred to as either masculine or feminine. These behaviors and characteristics are expressed through carriage (movement), dress, grooming, hairstyles, jewelry, mannerisms, physical characteristics, social interactions, and speech patterns (voice)'
Transgender	'An umbrella term for people whose gender identity and/or gender expression differs from their assigned sex at birth. May be abbreviated to trans'
Transgender man	'Generally refers to someone who was identified female at birth but who identifies and portrays his gender as male'
Transgender woman	'Generally refers to someone who was identified male at birth but who identifies and portrays her gender as female'
Cisgender	'People whose gender identity and gender expression align with their assigned sex at birth (i.e., the sex listed on their birth certificates)'
Gender non-conforming	'People whose gender expression is (1) neither masculine nor feminine or (2) different from traditional or stereotypic expectations of how a man or woman should appear or behave'

and new frontiers of research tend to amplify the experiences of individuals who identify as lesbian, gay, and bisexual, thereby focusing on sexual orientation rather than gender identity and gender fluidity (Alegria, 2011). This has led to underrepresentation of transgender and GNC individuals in research related to health care and, for the purpose of this study, evidence-based genetic practice recommendations.

A key component to genetic risk assessment and documentation of personal and family medical history is the genetics pedigree. As a visual representation of the genetic relationship between family members, a clinician can detect patterns of inheritance of genetic traits and features. Pedigree symbols and notations are standardized for easy and consistent communication among medical professionals and between the clinician and patient. For example, a circle denotes a female individual and a square represents a male (Bennett et al., 1995). An update in 2008 to standard pedigree recommendations by NSGC included nomenclature for a patient identifying as transgender (Bennett, French, Resta, & Doyle, 2008). The pedigree nomenclature suggests that a diamond can be for individuals whom their gender is not specified, with a difference of sexual development, or identifies as transgender. The authors note a circle with XY underneath could also be used to denote a transgender female and a square with XX could represent a transgender male.

In 2017, the National Comprehensive Cancer Network (NCCN) updated its nomenclature to include individuals who identify as 'transsexual' (transgender is the preferred term), suggesting they may be represented by a combination of symbols representing both their gender identity and biological sex. For example, a transgender

woman would be represented by a circle (representing her gender identity) with a square inside (representing her sex assigned at birth; Provenzale et al., 2017). Both the NCCN and NSGC make an impact on genetic counselors' practices, creating a potential for confusion among genetic counselors in having discordant recommendations. As with any other useful tool, periodic review of pedigree nomenclature is necessary to ensure that these standards are currently meeting the needs of the profession and are inclusive of all the patients we serve.

Much of the prior research in medicine concerning healthcare provision for the lesbian, gay, and bisexual (LGB) community focuses on provider preparedness. These studies found that providers were open to receiving more education on the subject and could use more training on cultural sensitivity to avoid heteronormativity (Kamen, Smith-Stoner, Heckler, Flannery, & Margolies, 2014). A further study not only confirmed this, but also established the overall lack of education surrounding LGB health care with 80% of nurses in the San Francisco Bay area reporting they had no education or training on LGB issues (Carabez et al., 2015). A survey from Glessner, Vandenlangenberg, Veach, and Leroy (2011) was sent to genetic counselors in order to assess their comfort level counseling LGB patients which was then compared to responses from patients on their experiences with genetic counseling. The authors found that counselors reported they would not alter their counseling approaches with their LGB patients while the patients expressed that the intake forms were not LGB friendly and they often had to correct heteronormative assumptions. These studies emphasize the need for more expansive and accessible LGB healthcare education.

Several sociological studies have shown that stigma against those who identify as transgender or GNC exists in the medical community and that this stigma can be very different than what LGB patients experience. This stigma can span from blatantly refusing to treat transgender or GNC patients to less direct forms of marginalization such as the absence of representation in medical intake forms (Cruz, 2014). The National Center for Transgender Equality surveyed those who identify as transgender on their healthcare experiences and found that 28% of respondents experienced outright harassment or violence in a medical setting and 19% were refused care (Grant et al., 2011). In a study conducted in 2011 assessing the National Transgender Discrimination Survey, at least 50% of respondents stated that they had to educate their healthcare providers in order to receive appropriate care (Grant et al., 2011). This takes time away from addressing patient concerns and treatment discussions and further marginalizes them. External experiences of stigma and discrimination can have a large impact on the individual's life, affecting their coping ability and well-being (Pryor, 2015). Cruz further asserts that making sure intake forms and family histories are inclusive to these patients can help to reduce that stigma little by little, increasing access to health care and hopefully the overall well-being of those who identify as transgender or GNC (Cruz, 2014).

As understanding of the marginalization of transgender and GNC individuals in multiple sectors of health care increases, the genetic counseling community will also need to increase awareness and cultural competency. Uncertainty surrounding best practices for pedigree nomenclature can perpetuate discomfort and hinder progress toward gaining cultural competency within the profession. A review of past sociological and therapeutic studies highlighted that acknowledgment and affirmation not only of who they are as they define themselves but also of their struggles has a positive effect on the overall well-being of the transgender individual (Connolly, 2005). In the practice of patient-focused counseling, genetic counselors are taught to affirm their patients own feelings toward genetic testing and the experiences that led them to feel that way (Uhlmann, Schuette, & Bashar, 2009). The purpose of this study was to identify the current practices regarding pedigree nomenclature genetic counselors use for patients identifying as transgender and GNC and report on education genetic counselors have both received and would like to receive regarding transgender and GNC health care. By examining these questions, the overall goal was to illuminate the variability in genetic counseling practice so that the profession can continue to be positioned at the forefront of providing culturally competent care for all patients.

## 2 | METHODS

### 2.1 | Participants

Members of NSGC received a recruitment notification through the NSGC's email blast to approximately 3,000 members (Appendix S1). The initial notification email included a description of the study,

participation requirements, and a link to the online survey. Four weeks after initial notification, a reminder email was sent including the same information. Anonymous responses were collected through Qualtrics software over a six-week period between November 2017 and January 2018.

Participants were required to either be a genetic counselor or genetic counseling student. The goal of the study was to include a broad spectrum of opinions from genetic counselors and genetic counseling students, and therefore, no exclusion criteria were applied with respect to practice area or work setting. The survey required participants to read and write in English. The study was reviewed by the Brandeis University Institutional Review Board and approved as human subjects' research.

### 2.2 | Instrumentation

The survey (Appendix S1) consisted of 36 questions and was designed to gather information in the following sections:

1. Demographics and background information.
2. History of educational and professional experiences focused on transgender and GNC medicine or gender identity and fluidity.
3. Confidence in ability to appropriately counsel transgender and GNC patients in a genetic counseling setting.
4. Current practice in pedigree nomenclature to represent transgender and GNC patients.
5. Genetics of risk assessment for case scenarios involving transgender patients with a family history of an X-linked condition.

Survey questions included a combination of multiple choice, Likert-scale, and open-ended responses. Sections 3 and 4 included hypothetical scenarios to allow participants to assess their own comfort levels in providing psychosocial counseling to patients who identify as transgender or GNC (data not included in this publication). The four scenarios involved a patient, identifying as a transgender man, transgender woman, gender non-conforming assigned female at birth, and gender non-conforming assigned male at birth, coming into the clinic for genetic counseling. There is no specific indication given as to why they are present for genetic counseling. In addition, participants were asked to choose a pedigree symbol from a list of options that they thought best represented their patient (Table 2). This list was intended to span the range from representing gender assigned at birth, gender identity, and incorporating both concepts in one symbol. Participants could also choose 'other' and further elaborate on their preferred designation if not represented on the list.

Prior to taking the survey, participants were provided with definitions for gender identity terms used in the survey (Glossary of Gender & Transgender Terms, 2010). The survey also included a link to an expanded list of terminology provided by Fenway Health to serve as a useful reference for the participants to access in the future.

**TABLE 2** Options of symbols participants could choose from when deciding which symbol they felt would best represent their hypothetical patient in the survey. The participants also had the option of selecting 'other' and describing what their ideal symbol would be, if they had one in mind

Scenario	Symbol	Rationale
Patient identifying as transgender male		'unspecified' as suggested by NSGC
		Representative of gender identity
		Representative of sex assigned at birth
		Representative of sex assigned at birth and gender identity, as suggested by NCCN
		Representative of sex assigned at birth and gender identity, as suggested by NSGC
		Representative of gender identity while acknowledging transition and not including karyotype information
Patient identifying as transgender female		'unspecified', suggested by NSGC
		Representative of sex assigned at birth
		Representative of gender identity
		Representative of sex assigned at birth and gender identity, suggested by NCCN
		Representative of sex assigned at birth and gender identity, suggested by NSGC
		Representative of gender identity while acknowledging transition and not including karyotype information

**TABLE 2** (Continued)

Scenario	Symbol	Rationale
Patient identifying as GNC – assigned male at birth		'unspecified', similarly suggested by NSGC for transgender patients
		Representative of sex assigned at birth
		Representative of gender identity as neither male nor female, while acknowledging likely karyotype based on sex assigned at birth
		Representative of 'unspecified' symbol with likely karyotype based on sex assigned at birth
Patient identifying as GNC – assigned female at birth		'unspecified', similarly suggested by NSGC for transgender patients
		Representative of sex assigned at birth
		Representative of gender identity as neither male nor female, while acknowledging likely karyotype based on sex assigned at birth
		Representative of 'unspecified' symbol with likely karyotype based on sex assigned at birth

**2.3 | Data analysis**

Statistical analyses were performed using IBM Statistical Package for Social Sciences (SPSS, version 24). We used demographic statistics to summarize demographic information. Chi-square analysis was used to assess associations between demographic information and symbol selection. Particular groups analyzed included whether or not they were practicing genetic counselors, practice area (distinguished between those who indicated oncology as part of their practice and those that did not), professional experiences with LGBTQ+ identifying individuals, and personal experiences with LGBTQ+ identifying individuals. McNemar analysis was performed to determine whether symbol selection varied significantly between the two scenarios involving transgender-identifying patients, as well as the two scenarios involving gender non-conforming patients. A  $p < .05$  was considered statistically significant.

Thematic analysis of open-ended responses was performed by the first author by using inductive content analysis to group

(Continues)

responses by their content (Elo & Kyngas, 2008). After reading through initial responses, six themes were identified and an Excel spreadsheet was used to determine which theme best represented each response in why the participant chose the symbol they felt best represented their patient. The six themes included clarity, genetic appropriateness, accuracy, alignment with guidelines, affirmation of patient gender identity, and feelings of uncertainty.

The entirety of this study was completed between May 2017 and May 2018.

### 3 | RESULTS

Four hundred ninety-eight participants began the survey, and 432 completed the survey in full. An additional 37 partially complete surveys were included in the analysis for a total of 469 responses.

#### 3.1 | Demographics and background information

The majority of the participants identified as cisgender female ( $n = 432/438$ , 98.6%), and 78% ( $n = 342/438$ ) reported being between 22 and 35 years of age. A majority ( $n = 240/438$ , 54.8%) had between 1 and 5 years of experience in their current practice area.

The current practice areas of respondents varied, with most ( $n = 300/438$ , 68.3%) in clinical practice, 10.3% ( $n = 45/438$ ) solely in Lab/Research, and 18.8% ( $n = 82/438$ ) were current students. Of those who currently practice patient-facing care, most ( $n = 240/438$ , 54.8%) reported seeing between 5 and 12 patients a week (Table 3).

When asked if participants had ever had a patient disclose as transgender or gender GNC, 61.2% ( $n = 287/469$ ) had said 'no' or 'unsure' while 38.8% ( $n = 182/469$ ) said yes. The majority of those asked if they personally knew someone who identified as transgender or GNC said yes ( $n = 258/468$ , 55.1%). Additionally, only 6.6% ( $n = 31/469$ ) of participants reported ever working in a clinic specifically serving patients from the LGBTQ+ community.

#### 3.2 | Preferences on pedigree symbols

Participants were first asked if they were aware of any nationally standardized recommendations of pedigree symbols representing transgender and GNC patients. Eighty-nine (out of 469, 19%) participants responded 'yes'. If participants responded 'yes', they were prompted to cite the source and a majority ( $n = 59/89$ , 66.3%) of those responses cited the Bennett et al. paper published in 2008 while 13.5% ( $n = 12/89$ ) cited the NCCN guidelines published in 2017. The remainder of participants listed that they had either heard about symbols from a NSGC Special Interest Group (SIG) discussion, discussed the topic in class during their training program, or relied on symbols more closely associated with genograms used in sociology.

Participants were then asked to select which pedigree symbol they felt best represented their patient out of a selection of symbols in four different scenarios. The four scenarios involved patients who identified as a transgender man, transgender woman, GNC patient assigned male at birth, and GNC patient assigned female at birth. No specific indication for the genetic counseling visit was given.

A total of 447 responses were recorded for both scenarios involving a patient identifying as transgender. 41.4% ( $n = 185/447$ ) of participants felt a symbol representing gender identity with a denotation of presumed genotypic sex assigned at birth based on external genitalia noted underneath would be most appropriate (Figures 1 and 2). The second most frequent option ( $n = 133/447$ , 29.7%) was a symbol denoting gender identity on the outside with the symbol denoting sex assigned at birth on the inside. There was no significant difference in a participant's selection when the scenario involved a patient identifying as a transgender man versus a transgender woman ( $p = .679$ ), indicating if a participant selects a particular symbol, they were likely to also chose a similar representative symbol in the following scenario. This was also the case in individual participants' selections for patients identifying as gender non-conforming ( $p = .488$ ).

We found that the distribution of selection of the seven presented options for symbols for patients identifying as transgender was statistically significantly associated with whether oncology was part of the participant's practice or not ( $\chi^2 = 17.215$ ,  $p = .004$ ,  $df = 6$ ). Specifically, those who disclosed that oncology was part of their practice were more likely to select the symbol with one nested within another to show gender identity and sex assigned at birth (87/153 [56.8%], 70/285 [24.6%], respectively). No significant differences were observed for the following groups: currently practicing versus not, have ever practiced versus students, professional experience (either in a specific clinic or patient disclosure) with LGBTQ+ individuals versus no professional experience with LGBTQ+ individuals, and personal experience with LGBTQ+ individuals versus no personal experience with LGBTQ+ individuals ( $p > .05$  for all analyzed groups' symbol selections).

For those that chose an 'other' symbol, some additional suggestions included noting 'MTF' or 'FTM' under a symbol representing their preferred gender, indicating Male to Female or Female to Male, respectively. Others responded that it would be best to use the symbol according to sex assigned at birth and notate the patient's gender identity underneath.

When asked to explain why participants felt their selection would be most appropriate for their transgender patient, many participants responded with the same themes, even with having chosen different symbols. These common themes focused on clarity, genetic appropriateness, accuracy, aligning with guidelines, and affirmation. Some who selected a symbol in-line with NSGC or NCCN guidelines had cited those guidelines as the rationale for their choice; however, a majority of participants were not aware of those guidelines, so other rationales became apparent as well. One participant who chose a circle with a square inside as the most appropriate symbol for a transgender woman explained that this selection was most clear:

**TABLE 3** Summary of key demographic information obtained in this study. LGBTQ+ stands for lesbian, gay, bisexual, transgender, queer, and others. GNC stands for gender non-conforming

Variable	n	%
Age		
<22	1	0.23
22-35	342	78.08
36-49	68	15.53
50-63	26	5.94
64-70	1	0.23
>70	0	0
Total	438	100
Years of patient-facing care		
1-5	240	54.80
6-10	55	12.56
11-15	22	5.02
16-20	15	3.42
20+	24	5.48
I'm currently a student	82	18.72
Total	438	100
Current practice field (Can pick more than one, total n = 438)		
Oncology	153	34.93
Pediatrics	99	22.6
Prenatal	107	24.43
Adult/general	65	14.84
Other specialty clinic	51	11.64
Lab/research/other	64	14.61
Years in current practice field		
1-5	255	58.36
6-10	50	11.44
11-15	19	4.35
16-20	16	3.66
20+	15	3.43
I'm currently a student	82	18.76
Total	437	100.00
Patients per week		
0-4	104	24.07
5-8	133	30.78
9-2	113	26.16
13-16	51	11.81
More than 16	31	7.18
Total	432	100.00
Worked in an LGBTQ+ clinic		
Yes	31	6.61
No	438	93.39
Total	469	100
Had a patient disclose they are transgender or GNC		

(Continues)

**TABLE 3** (Continued)

Variable	n	%
Yes	182	38.81
No	270	57.57
Unsure	17	3.62
Total	469	100
Personally know someone who identifies as transgender or GNC		
Yes	258	55.13
No	210	44.87
Total	468	100

*This represents that there will be physiologic, anatomic and psychosocial differences from a cis-gender female*

Other participants cited that their symbol choice which did not include karyotypic information was the most appropriate to keep the pedigree accurate. This participant chose a square with the denotation 'trans male' underneath for a transgender man and wrote:

*I don't know their chromosome complement [sic] so I can't write XY or XX.*

Some felt genetics took precedent in order to give an accurate pedigree for outside interpretation and chose symbols solely representing sex assigned at birth:

*For the sake of a genetics pedigree I think we have to honor and be consistent in documenting the biological genetic makeup of all individuals (especially in regards to x-linked traits, or conditions that more commonly manifest themselves in biologic men or women, etc.)*

Overwhelmingly, participants wanted to affirm their patients' identities and struggles and felt that a pedigree was one way in which to do so. One participant even went as far as to reflect on their own selection and making sure they were truly affirming the patient's experiences:

*She does identify as female so in my own work I usually use the square inside a circle as it makes the most sense to me (though perhaps I should reflect on why I see such patients as a gender other than female... what I've been using implies female on the outside, male on the inside, and I think that is actually problematic, now that I'm thinking deeply about it!).*

In both scenarios involving a patient identifying as GNC, a diamond with the denotation GNC and their assumed genotypic sex underneath was most frequently selected ( $n = 200/438$ , 45.6%) as the best symbol to represent their patient (Figures 3 and 4). Additionally, more participants than in the case with a transgender patient felt it would be appropriate to use a diamond or the symbol representing

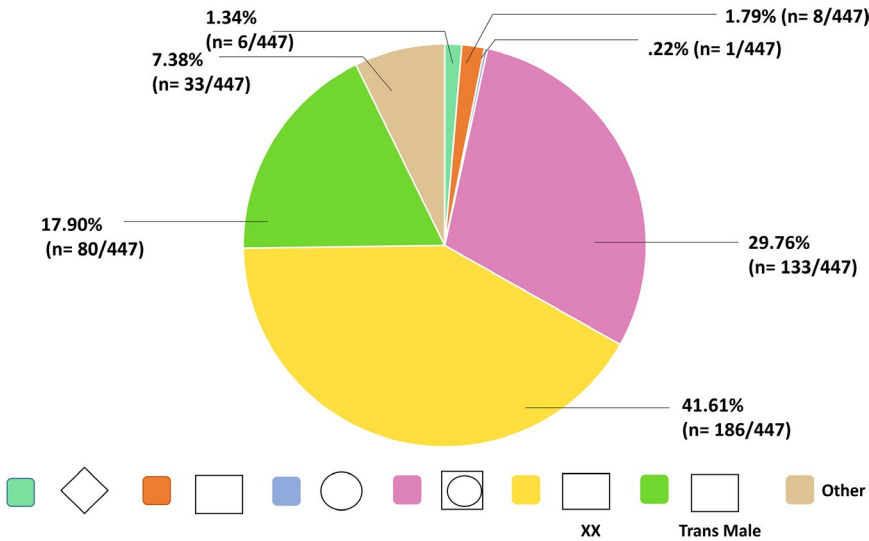


FIGURE 1 Breakdown of the frequency in which symbols were chosen to represent transgender male patients

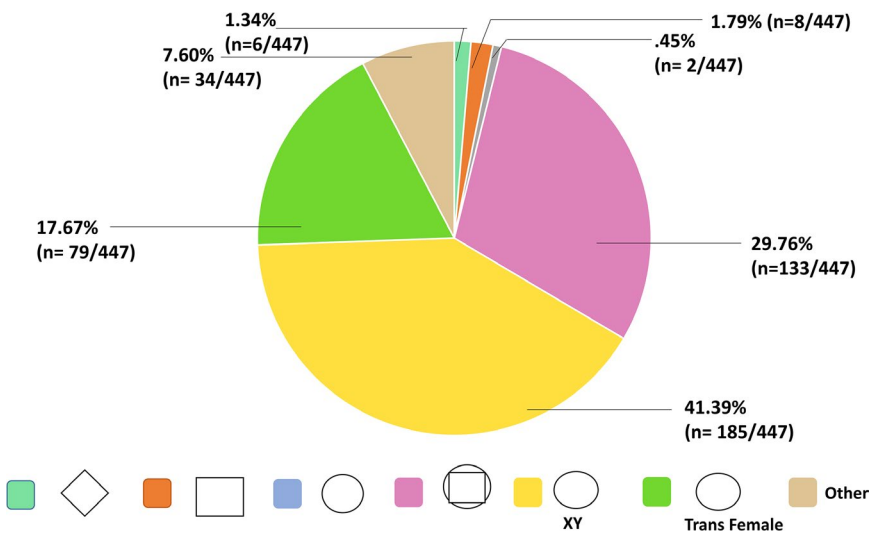


FIGURE 2 Breakdown of the frequency in which symbols were chosen to represent transgender female patients

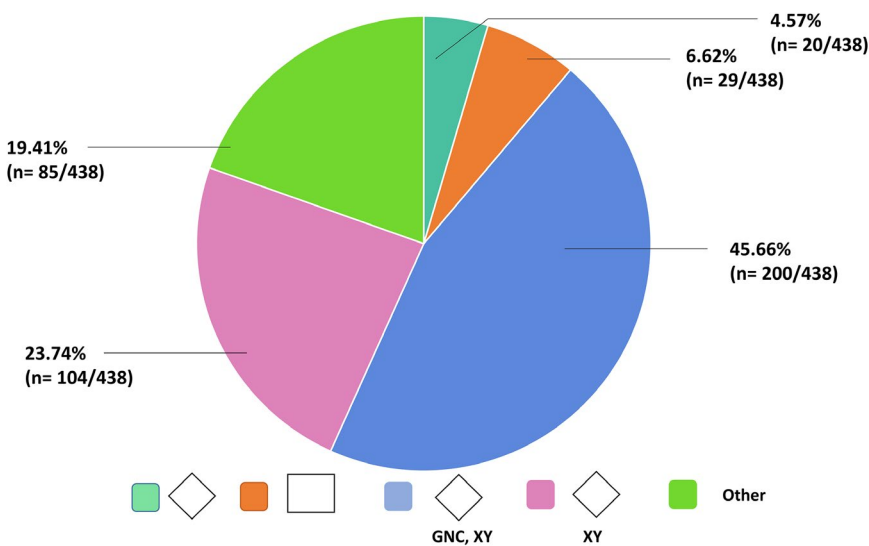
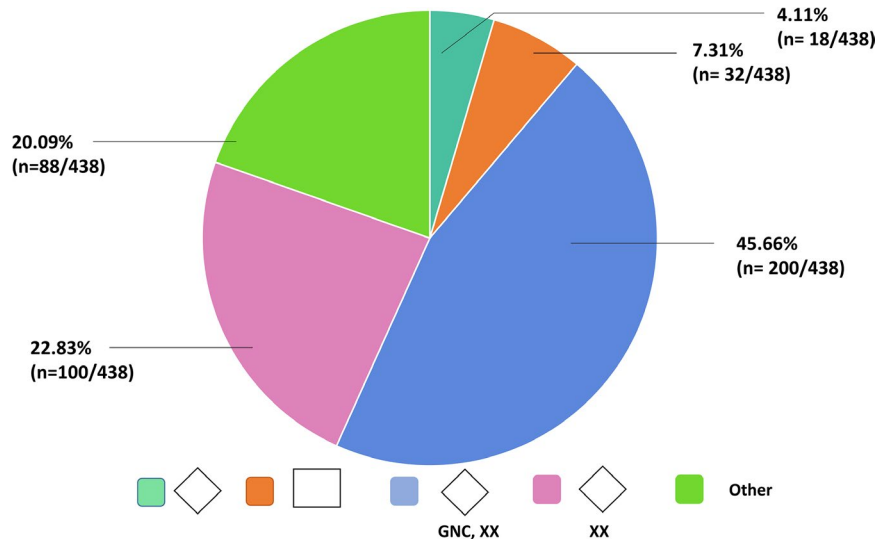


FIGURE 3 Breakdown of the frequency in which symbols were chosen to represent gender non-conforming (GNC) patients assigned male at birth (AMAB)

sex assigned at birth without any further denotation. There was no significant association between nomenclature for patients identifying as GNC and the four groups analyzed (Experienced genetic

counselors compared with students, those who indicated oncology as part of their practice and those that did not, professional experiences with LGBTQ+ individuals compared with those who do not,

**FIGURE 4** Breakdown of the frequency in which symbols were chosen to represent gender non-conforming (GNC) patients assigned male at birth (AFAB)



and personal experiences with LGBTQ+ individuals and those without any) ( $p > .05$ ).

Participants who chose an 'other' symbol, many suggested a symbol that was more consistent with some of the symbols available for transgender patients. A few of the participants thought there should be an option of a diamond with a smaller circle or square (denoting sex assigned at birth) while others felt a diamond with the notation AFAB or AMAB (for assigned female at birth and assigned male at birth, respectively) underneath would be appropriate. Another area of uncertainty among counselors was language. Some were concerned that while a symbol may have made sense to them, the terms associated with the transgender and GNC communities may not be common knowledge.

*I'm not sure that "GNC" is an acronym recognized by most GCs, though I would certainly support using "GNC" as a designator.*

*I stumble a bit with gender neutral pronouns (they/their).*

When asked to explain why participants felt their selection would be most appropriate for their GNC patient, some of the same themes of making sure symbols were clear, accurate, and affirming appeared. A participant who chose the diamond with GNC and their sex assigned at birth underneath explained:

*This best represents who they are; it's like why I include adopted children in a pedigree even though they aren't biologically related - they are still family and have a context in that patient's life like a person's gender does.*

Participants again felt that a pedigree should be genetically accurate and felt uncomfortable listing an assumed karyotype without performing one. They reported being overall unsure of what the most appropriate symbol would be. Of the available options, many felt that what they chose was consistent with what they felt was most appropriate for their transgender patients:

*I do not know it is the best choice, but I would have put a square inside of a diamond to follow the trend of the other standard symbols. I believe this is the best because the patient has not had a karyotype done (so we don't know they are actually XY) but it still affirms their gender.*

A new theme of uncertainty arose within these scenarios. A participant responded with complete uncertainty, welcoming a new choice altogether:

*I don't feel that any of these are appropriate, but I don't have a better idea either...I think maybe an entirely new symbol would be best*

### 3.3 | Education on transgender and GNC health care

Participants were asked what education they have received regarding transgender and GNC health care and were given 6 options, of which they could choose multiple. The options included 'I have attended or listened to lectures', 'I have sought out my own sources on the internet', 'I have read scholarly articles or books on the topic', 'I have reached out to colleagues who may be more knowledgeable', 'I have attended a workshop', and 'I have not had any specific education on this topic'. A total of 437 participants responded and 81.5% ( $n = 356/437$ ) sought education on the topic, with attending lectures and seeking resources on the internet being the most common choice (Figure 5a).

Participants were then asked what educational resources they would like to have regarding transgender and GNC health care and were again given six options, of which they could choose multiple. The options included 'online learning modules or webinars', 'lectures', 'workshops', 'more research', 'books or pamphlets', and 'nothing, the education is sufficient'. The overwhelming majority ( $n = 431/437$ , 98.6%) reported a desire for more education, with 'online learning modules or webinars' receiving the most responses (Figure 5b).



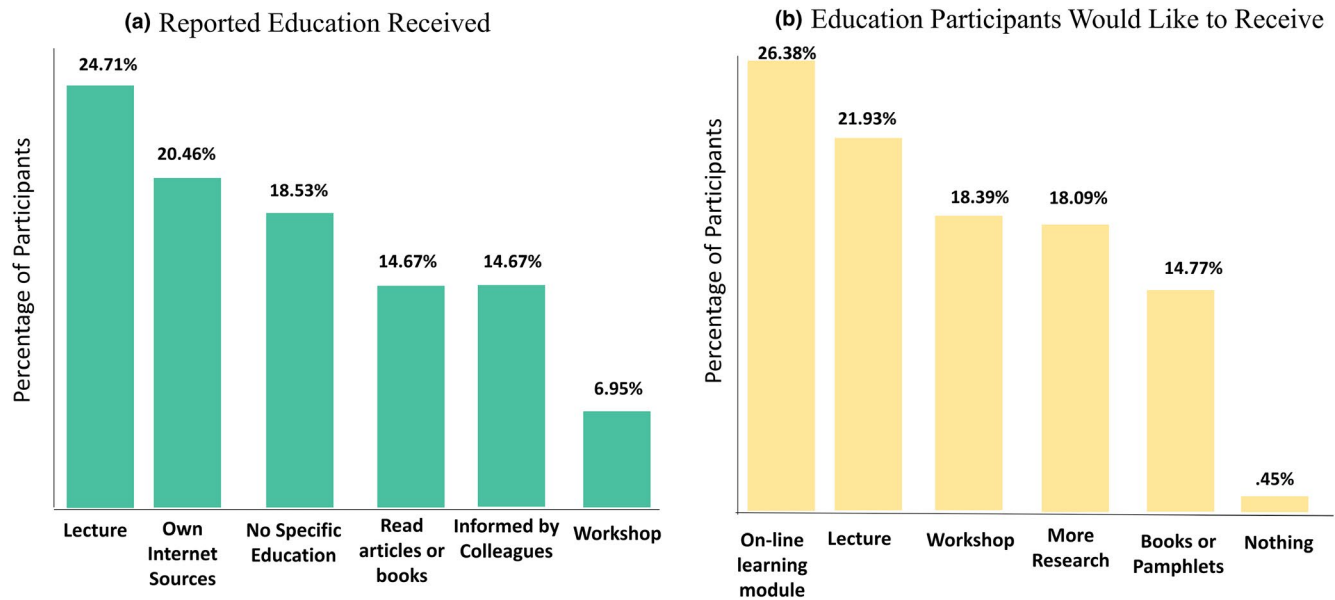


FIGURE 5 Education participants have had and would like to see in transgender and gender non-conforming health care

#### 4 | DISCUSSION

This study served to assess current practices in genetic counseling for transgender and GNC patients. Very few genetic counselors reported having had professional experience counseling transgender and GNC patients, though a greater proportion reported that they have personal connections with these communities. The education participants have received on transgender and GNC health care varied; however, most agree that there needs to be more education on the topic. Genetic counselors felt more than one pedigree symbol could be appropriate in these sessions and their choice was driven by a desire to affirm their patients' experiences. A recent survey found that genetic counselors in their study were uncomfortable asking patients about their pronouns, which presents an opportunity for targeted education (Berro, Zayhowski, Field, Channaoui, & Sotelo, 2019).

Within the need for more education, genetic counselors also reported wanting more guidance with regard to pedigree nomenclature for their transgender and GNC individuals. The two most favored symbols for representing transgender patients reflected the recommendations set forth by NSGC and NCCN. The third most commonly chosen option similarly recognized gender identity as the primary symbol, but included more descriptive information underneath. Many variables were analyzed to determine whether factors such as years of experience, age, and gender identity had any impact on nomenclature selection. The only association of significance identified was that those who practiced oncology were more likely to select a symbol more in-line with NCCN guidelines, whereas every other practice area was more likely to select a symbol more in-line with NSGC guidelines or another symbol. This was a particularly interesting observation given 81% of participants report being unaware of standardized nomenclature.

When it came to selecting a symbol for GNC patients, the majority felt that a diamond, as recommended by NSGC, with some

adjustments would be most appropriate. Many respondents stressed that if a diamond is chosen to represent an individual who identifies as GNC that is simultaneously important to notate 'GNC' so that in the absence of notation the diamond is not confused with 'gender unknown'. A greater number of participants selected 'other' in response to the question of pedigree symbols for GNC patients compared with transgender individuals, with many having stated that they welcomed a new symbol that would be unique for GNC patients since they do not fit within the gender binary. Our findings suggest a high degree of discordance within the genetic counseling community on pedigree nomenclature for GNC individuals. No demographic information, including practice area, had an impact on symbol selection, further differentiating this data from the data collected using scenarios involving a transgender male or female patient.

More important than the symbols chosen were the rationales given for each choice. Genetic counselors reported feeling that the choice they selected was most appropriate and most accurately represented their patients and this same justification was observed in each of the symbol options. Affirmation of client gender identity was particularly important, speaking to the client-centered approach of genetic counseling (Uhlmann et al., 2009). In the same spirit that adopted children are included in pedigrees, many counselors felt that when drawing a pedigree in front of a patient it is important to make an accurate representation not only of their genetic family, but of their gender identity (Bennett et al., 2008). The importance of affirmation and inclusivity has been demonstrated in a recent qualitative study in which the researchers engaged members of the transgender and gender non-binary community on their opinions of three different pedigree symbol options and found that most felt that a single circle, square, or diamond to denote gender identity followed by annotation of sex assigned at birth to be most clear and inclusive (Barnes, Morris, & Austin, 2019). Furthermore, participants asserted that the onus falls on genetic counselors to create an environment of

safety and explain why sex assigned at birth and gender identity are important to the clinical setting.

While affirmation is important, many genetic counselors also felt that a pedigree needs to be genetically accurate, making sure that there is information distinguishing gender identity and sex assigned at birth. This makes sense, as it needs to be easily understood by many health professionals who use a pedigree as a tool to determine the best care for the patient. Along those lines, many felt strongly that karyotype information, such as denoting XX or XY at the bottom of a symbol, should not be included unless one has been obtained. Some offered that a notation underneath such as MTF (male to female), FTM (female to male), or GNC would be a better option than a karyotype.

The discrepancy in what should be the best symbol to use in each scenario highlights an important point of uncertainty within the genetic counseling community, which may impact other healthcare providers who rely on the pedigree standardization for interpretation and genetic risk assessment. Regardless of the symbol chosen, participants cited that their selection was the best choice because of its accuracy and clarity, which means that different symbols are more clear to some than others. A potential contributing factor to this is the fact that only 19% of participants cited that they were aware of national standardized recommendations and of those that did, they cited two different organizations with two different suggestions for appropriate symbols.

One topic genetic counselors agreed on in this survey was the desire for more education on transgender and GNC health care. A qualitative study recently published documents feelings of unpreparedness for counseling sessions with transgender patients (Zayhowski et al., 2019). Genetic counselors in our study widely supported more education through readily accessible formats such as online webinars or lectures, workshops, and new research, which demonstrates a recognition that increased self-education as an effective strategy for increasing awareness and improving competency. Workshop formats can promote engagement and focus on addressing concerns regarding the use of gender-inclusive language and LGBTQ+ cultural competency for genetic counselors and other healthcare providers (Chisolm-Straker et al., 2017; Glessner et al., 2011).

Workshops at both national and regional conferences could address some of the inconsistencies evidenced by these data. People may not be aware of how ill-prepared they are to address a clinical situation until it is presented to them in clinic, ultimately impacting the patient's experience and counselors' alliance with them. If genetic counselors were able to question their own preparedness and receive guidance from those with more experience, they can ensure bringing a higher level of understanding to future encounters.

#### 4.1 | Study limitations

One of the limitations of the study was that it was only made available to genetic counselors and students who receive emails from

NSGC's listserv. While we were able to gather many responses across different specialties and broad years of experience, it still was not inclusive of all genetic counselors and students who may not be part of NSGC or who did not participate in the survey. Even though the study did note discrepancies between those who used NSGC versus NCCN guidelines for pedigree nomenclature, there may have been an underrepresentation of those who would have followed the NCCN guidelines because the survey was only sent through the NSGC listserv. An additional limitation was that the scope of the study was limited to questions aimed at understanding current practices for transgender and gender non-conforming symbols and did not include questions directed at current practices for representing intersex patients.

#### 4.2 | Research recommendations

The results of this study reveal two key areas for further investigation. First, the data show variability in pedigree nomenclature in current practice among genetic counselors and students when considering affirming symbology for transgender and GNC patients. More expansive research, including more pedigree interpretation involving patients identifying as transgender and GNC, would demonstrate counselors' and other healthcare professionals' abilities to interpret familial risk in the absence of universally standardized nomenclature. A second area of research could focus on the impact of further education on the lived experiences of individuals with transgender and GNC identities on genetic counseling competency. Although our study revealed a desire for more education, more studies are needed on the efficacy of different educational approaches for genetic counselors and trainees. Any educational opportunities should be created in partnership with LGBTQ+ healthcare advocacy groups to reduce the risk of reinforcing stereotypes and improve knowledge. It is very clear that patients identifying as transgender and gender non-conforming feel they still have to educate their healthcare professionals regarding the needs of their care, which creates an imbalance for the patient-provider relationship (Grant et al., 2011). We also acknowledge that our study did not include data identifying preferred pedigree nomenclature and education surrounding patients identifying as intersex. Equitable care for all gender minority groups relies on future research to consider topics that pertain to all of those who identify as a gender minority.

#### 4.3 | Practice implications

While this study demonstrates the variation among genetic counselors as it pertains to appropriate symbol use for patients identifying as transgender and GNC, it also illustrates what needs to be considered when establishing standardized pedigree nomenclature. A key advantage to standardized nomenclature is consistency in interpretation of the family structure in risk assessment by healthcare

providers (Bennett et al., 1995, 2008; Provenzale et al., 2017). One of the primary highlights from this study is that genetic counselors have varying ideas as to what symbols are most appropriate for their patients, with use of NCCN or NSGC recommendations associated with practice area. While considering how to best update standardized nomenclature to be more gender-inclusive, it may be helpful to have multiple genetics professional organizations come together to make a joint statement in order to limit discrepancies in pedigree creation and interpretation. Regardless of symbol selection, participants overwhelmingly wanted to pursue further education to better understand their patients' experiences and gender identity. Professional organizations can build upon this well-meaning spirit to spearhead more gender-competent trainings, focusing on language, cultural humility, and affirming treatment.

## 5 | CONCLUSIONS

Over 400 genetic counselors and trainees participated in this study. Their responses illuminated the variability in current practices in pedigree nomenclature for transgender and gender non-conforming patients. Genetic counselors questioned their level of preparedness in caring for patients in an affirming way, which is central to the client-centered approach to genetic counseling. A core value within the profession is providing client-centered care, and the basis for a trusting alliance in a session can be demonstrated through the skills and tools used to obtain and record information about their patients. These findings demonstrate that the genetic counseling community has not come to a consensus regarding appropriate nomenclature that would be most easily interpreted and representative of their transgender or GNC patient on a pedigree. Such variability within current practice could inhibit progress toward providing culturally competent care. Confusion and uncertainty on the interpersonal level are likely to hinder client-centered care; thus, working toward consistency and competency within the profession will contribute to promoting the best possible care for transgender and GNC individuals seeking genetic counseling services.

### AUTHORS CONTRIBUTIONS

**Elizabeth Sheehan:** Ms. Sheehan conceived of this project, involved in project design, collected, analyzed, and interpreted data, made significant contributions to the drafting of the work, provided final approval for the version to be published, and agreed to be accountable for all aspects of the work regarding accuracy and integrity. **Gayun Chan-Smutko:** Ms. Chan-Smutko supervised this project from its conception, involved in project conception and design, interpreted data, made significant contributions to the drafting of the work, provided final approval for the version to be published, and agreed to be accountable for all aspects of the work regarding accuracy and integrity. **Robin Bennett and Miles Harris:** Dr. Bennett and Mr. Harris were involved in survey design, made revisions to the manuscript, provided final approval for the version to be published, and agreed to be accountable for all aspects of the work regarding accuracy and integrity.

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### COMPLIANCE WITH ETHICAL STANDARDS

#### Conflicts of interest

Elizabeth Sheehan, Gayun Chan-Smutko, Robin Bennett, and Miles Harris declare that they have no conflict of interest.

#### Human studies and informed consent

This study was reviewed by the Brandeis University Institutional Review Board and approved as human subjects' research. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all participants for being included in the study.

#### Animal studies

No non-human animal studies were carried out by the authors of this article.

### DATA AVAILABILITY STATEMENT

Data used for the analyses of this publication have not, to date, been archived in a public repository. Data can be made available upon request by contacting the first author.

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### SUPPORTING INFORMATION

Additional supporting information may be found online in the Supporting Information section.

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