Geometric Inclusivity: An Assessment of Current Practices in Pedigree Nomenclature for Patients Identifying as Transgender and Gender Nonconforming

Master’s Thesis

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Gayun Chan-Smutko, Advisor

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Master of Science
in
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by
Elizabeth Sheehan

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Abstract

Geometric Inclusivity: An Assessment of Current Practices in Pedigree Nomenclature for Patients Identifying as Transgender and Gender Nonconforming

A thesis presented to the Graduate Program in Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

By Elizabeth Sheehan, MS, GC

The healthcare community is attempting to expand its knowledge of LGBT health. Much of the existing literature and research amplifies the experiences of individuals who identify as lesbian, gay, and bisexual, with less focus specifically on transgender and gender nonconforming (GNC) health. One potential barrier to uniform representation and consistent care for patients who identify as transgender or GNC is the inconsistency of accepted pedigree symbols to represent them. Both the National Society of Genetic Counselors and National Comprehensive Cancer Network have differing guidelines for what are acceptable pedigree symbols to represent transgender patients and minimal recommendations for GNC patients. We surveyed genetic counselors to explore current practices of pedigree nomenclature for patients identifying as transgender and GNC and their level of confidence in addressing their psychosocial needs. Genetic counselors reported that using pedigree nomenclature that represented an individual’s gender identity was important; however, there was no consensus on best practice. This study also revealed a desire and need for more education and highlighted how education can positively
influence a genetic counselor’s confidence in addressing their transgender and GNC patient’s psychosocial needs.

Keywords: pedigree symbols, transgender, gender nonconforming, genetic counseling, education, confidence
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Introduction

The population of patients identifying as LGBT (lesbian, gay, bisexual, and transgender) to their healthcare providers has increased over the last decade, prompting healthcare providers to seek out additional resources outside of their traditional training to provide adequate care for these patients (Fredriksen-Goldsen et al., 2014). The healthcare community is attempting to expand its knowledge of LGBT health; however, the experiences of transgender and gender nonconforming (GNC) individuals are often rendered invisible under the broad LGBT umbrella. The transgender and GNC community itself represents a complex and diverse set of identities and terminology used within the community can differ and evolve over time. For this study, we referred to the definitions offered by Fenway Health (Glossary of Gender and Transgender Terms, 2010) in which transgender is a “an umbrella term used to represent those whose gender identity and/or gender expression differs from their assigned sex at birth”. Gender nonconforming is a term for “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”. Much of the existing literature 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 an imbalance of inclusion of transgender and GNC individuals in many studies related to healthcare and, for the purpose of this study, genetics practices. The dearth of medical genetics and genetic counseling publications on genetics services for transgender and GNC
patients further underscores the lack of evidence-based recommendations for culturally competent care of this population.

A key component to genetic risk assessment and documentation of personal and family medical history is the genetics pedigree. A pedigree is a visual representation of degree of relationship between family members. By recording pertinent family medical history, a clinician can detect patterns of inheritance of genetic traits and features. Pedigree symbols and notations are standardized for easy 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 the National Society of Genetic Counselors (NSGC) included nomenclature for sperm and egg donors (Bennett et al, 2008). The pedigree nomenclature suggests that a diamond can be for individuals whom their gender is not specified. A diamond can also be used if a patient has a sex development disorder or is transgender. The authors note that a circle with XY underneath could also be used to denote a transgender female and a square with XX underneath could represent a transgender male.

In 2017, the National Comprehensive Cancer Network (NCCN) updated their 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 women 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 in 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 is inclusive of all of 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 et al., 2014). A further study not only confirmed this, but also established the overall lack of education surrounding LGB healthcare with 80% of the 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 et al. 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 (Glessner et al., 2011). These studies in particular emphasize the need for more expansive and accessible LGBT healthcare education.

More sociological studies have shown that stigma against those who identify as transgender or gender nonconforming exists in the medical community and can span from blatantly refusing to treat them to less direct forms of marginalization such as the absence of representation in medical intake forms (Cruz, 2014). 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 that intake forms and family histories are inclusive to these patients can help to reduce that stigma little by little, increasing
access to healthcare and hopefully the overall wellbeing of those who identify as transgender or nonconforming (Cruz, 2014).

Examples of concrete steps towards reducing health disparities are evident, such as in the development of a LGBT Cancer Action Plan to address the psychosocial needs of LGBT patients which are different from the needs of cis-gendered heterosexual patients (Burkhalter et al., 2016). On a larger scale, Fredriksen-Goldsen et al. proposed expanding the civil rights act to include gender and sexual identity in order to combat institutionalized discrimination against LGBT people (Fredriksen-Goldsen et al., 2014). The field of medical genetics and genetic counseling is not immune to the need to identify and address the specific needs of LGBT patients.

As understanding of the marginalization of transgender and GNC individuals in multiple sectors of healthcare increases, the genetic counseling community will also need to increase awareness and cultural competency skills. Uncertainty surrounding best practices for pedigree nomenclature can perpetuate discomfort and hinder progress towards gaining cultural competency within the profession as a whole. The purpose of this study was to 1) identify the current practices regarding pedigree nomenclature genetic counselors use for patients identifying as transgender and GNC, 2) assess confidence in addressing the psychosocial needs of transgender and GNC patients, and 3) report on education genetic counselors have both received and would like to receive regarding transgender and GNC healthcare. By examining these questions, the overall goal was to illuminate the variability in genetic counseling practice so that the profession can continued to be positioned in the forefront of providing culturally competent care.
Methods

Recruitment

Members of NSGC received a recruitment notification through the NSGC’s email blast to approximately 3,000 members (Appendix A). 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. The anonymous survey was anonymous and made available through Qualtrics software for six weeks. Participants consented to participate in the survey by clicking through to the survey and selecting “I consent to taking this survey” on the first page, which included the description of the study.

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. At the end of the survey, participants were offered the opportunity to enter a random drawing for one of three $50 Amazon gift cards. The study was approved by the Brandeis University Institutional Review Board.

Instrument Design

The survey (Appendix B) 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 gender nonconforming medicine or gender identity and fluidity.

3. Confidence in ability to appropriately counsel transgender and gender nonconforming patients in a genetic counseling setting.

4. Current practice in pedigree nomenclature to represent transgender and gender nonconforming 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 combinations of multiple choice, Likert-scale, and open-ended responses. Section 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 gender nonconforming. In addition, participants were asked to choose a pedigree symbol from a list of options which they thought best represented their patient. 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.

Section 5 included two scenarios involving transgender patients who come in for genetic counseling regarding a family history of Duchenne Muscular Dystrophy, an x-linked condition. Participants were asked to assess the risk to a future child of the patient given the family history and the patient’s personal medical history.

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

**Data Analysis**

Data analysis included frequencies, descriptive statistics, correlation analyses, and regression analysis using SPSS software version 24. Thematic analysis was used for open-ended responses.

In order to determine if demographics and past experience with LGBT, specifically transgender and GNC, individuals impacted symbol selection and confidence level, independent sample t-tests were used to compare confidence, pedigree selection, and education based on demographic data. Regression analysis was performed in order to establish whether there were any further correlations and significance for those same values. Correlations were performed using one-way ANOVA Bonferroni analysis and chi square.
Results

Four hundred ninety-eight participants began responded to 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.

Demographics and background information

The majority of the participants (n = 438) identified as cisgender female (92.6%), while 6.4% identified as cisgender male. Two participants identified as GNC and none identified as either transgender male or female. Two preferred not to disclose. Seventy-eight percent reported being between 22 and 35 years of age and a majority (58.8%) had between 1-5 years of experience in their current practice area.

The current practice areas of respondents varied between Oncology (24.6%), Pediatrics (15.9%), Prenatal (17.2%), Adult/General (10.5%), Lab/Research (10.3%) and other specialties or fields (8.2%). Of those who currently practice patient-facing care, most (56.9%) report seeing between 5 and 12 patients a week (n = 432). Eighteen percent of respondents reported that they are a student in a genetic counseling training program (Table 1).

When asked if participants had ever had a patient disclose as transgender or gender nonconforming (GNC), 61.2% had said “no” or “unsure” while 38.8% said yes. When asked if they personally knew someone who identified as transgender or GNC, the majority (55.1%) said yes. Additionally, 6.6% of participants reported ever working in a clinic serving specifically patients from the LGBT community (Table 2).
### Table 1: General Demographic Information of Participants

<table>
<thead>
<tr>
<th>Variable</th>
<th>n</th>
<th>%</th>
<th>Mean</th>
<th>Range</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Age</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>&lt;22</td>
<td>1</td>
<td>0.23%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>22 - 35</td>
<td>342</td>
<td>78.08%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>36 - 49</td>
<td>68</td>
<td>15.53%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>50 - 63</td>
<td>26</td>
<td>5.94%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>64 - 70</td>
<td>1</td>
<td>0.23%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>&gt; 70</td>
<td>0</td>
<td>0%</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td>438</td>
<td>100%</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Years of patient-facing care</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>1 - 5</td>
<td>240</td>
<td>58.35%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>6 - 10.</td>
<td>56</td>
<td>11.44%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>11 - 15</td>
<td>22</td>
<td>4.35%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>16 - 20</td>
<td>15</td>
<td>3.66%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>20+</td>
<td>24</td>
<td>3.43%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>I'm currently a student</td>
<td>82</td>
<td>18.76%</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td>438</td>
<td>100%</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Current practice field (Can pick more than one, total participants = 438)</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Oncology</td>
<td>153</td>
<td>24.64%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Pediatrics</td>
<td>99</td>
<td>15.94%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Prenatal</td>
<td>107</td>
<td>17.23%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Adult/General</td>
<td>65</td>
<td>10.47%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other specialty clinic</td>
<td>51</td>
<td>8.21%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Lab/Research/Other</td>
<td>64</td>
<td>10.31%</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Years in current practice field</strong></td>
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<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>1 - 5</td>
<td>255</td>
<td>58.35%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>6 - 10.</td>
<td>50</td>
<td>11.44%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>11 - 15</td>
<td>19</td>
<td>4.35%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>16 - 20</td>
<td>16</td>
<td>3.66%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>20+</td>
<td>15</td>
<td>3.43%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>I'm currently a student</td>
<td>82</td>
<td>18.76%</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td>437</td>
<td>100.00%</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Patients Per Week</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>0 - 4</td>
<td>104</td>
<td>24.07%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>5 - 8</td>
<td>133</td>
<td>30.79%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>9 - 12</td>
<td>113</td>
<td>26.16%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>13 - 16</td>
<td>51</td>
<td>11.81%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>More than 16</td>
<td>31</td>
<td>7.18%</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td>432</td>
<td>100.00%</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>% of time spent on clinical activities</strong></td>
<td>48.50%</td>
<td>0 - 100%</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

### Table 2: Participants’ experiences with LGBT, specifically transgender and GNC, patients

<table>
<thead>
<tr>
<th>Variable</th>
<th>n</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Worked in an LGBT clinic</td>
<td>Yes</td>
<td>31</td>
</tr>
<tr>
<td></td>
<td>No</td>
<td>438</td>
</tr>
<tr>
<td>Had a patient disclose they are trans or GNC</td>
<td>Yes</td>
<td>182</td>
</tr>
<tr>
<td></td>
<td>No</td>
<td>270</td>
</tr>
<tr>
<td></td>
<td>Unsure</td>
<td>17</td>
</tr>
<tr>
<td>Personally know someone who identifies as trans or GNC</td>
<td>Yes</td>
<td>258</td>
</tr>
<tr>
<td></td>
<td>No</td>
<td>210</td>
</tr>
</tbody>
</table>
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. 86 participants responded “yes”. If participants responded “yes”, they were prompted to cite the source and a majority (69.1%) of those responses cited the Bennet et al. paper published in 2008 while 14.2% cited the NCCN guidelines published in 2017. The remainder of participants listed that they had either heard about symbols from a 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. These scenarios involved patients coming in for genetic counseling that identified as a transgender man, transgender woman, GNC assigned male at birth, and GNC assigned female at birth. No specific indication for the visit was given.

Four hundred and forty-seven participants’ responses were recorded for both scenarios involving a patient identifying as transgender and the majority of participants felt a symbol representing gender identity with a denotation of genetic sex assigned at birth noted underneath would be most appropriate (Figure 1A and B). The second most frequent option (29.7%) was a symbol denoting gender identity on the outside with the symbol denoting sex assigned at birth on the inside. A symbol representing solely gender identity was chosen by only 2 participants. There was no significant difference in the participant’s selection when the scenario involved a patient identifying as a transgender man versus a transgender woman. Those who disclosed that they work in Oncology were significantly (p = .004) more likely to select the symbol with one nested
within another to show gender identity and sex assigned at birth compared to those who worked in other specialties.

**Figure 1: Breakdown of the frequency in which symbols were chosen transgender patients**

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. Others responded that it would be best to use the symbol according to genetic sex, but 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. Many felt their choice was the clearest or most accurate representation of their patients. A participant who chose a circle with a square inside as the most appropriate symbol for a transgender woman explained that:

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

Additionally, other participants cited that their choice, usually in combination with a symbol choice that did not include karyotype, was the most genetically appropriate. For example, this
participant chose a square with the denotation “trans male” underneath for a transgender man and included their rationale:

“I don't know their chromosome compliment so I can't write XY or XX.”

Others felt genetics took precedent in order to give an accurate pedigree to those who may need to interpret it at another point:

“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.)”

Overall, participants wanted to be able 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, the majority of participants (45.6%) felt a diamond with the denotation GNC and their genotypic sex underneath would best represent their patient (Figure 2A and B) (n = 438). Additionally, more participants than in the case with a transgender patient felt it would be appropriate to use a diamond or the symbol representing sex assigned at birth without any further denotation. There was no significant difference in choices between a patient identifying as GNC and assigned male at birth versus female at birth.

For those that chose an “other” symbol, which was at a higher frequency than in the case of having a transgender patient, 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 sufficient.

**Figure 2:** Breakdown of the frequency in which symbols were chosen for GNC patients

<table>
<thead>
<tr>
<th>Pedigree Symbols for Gender Nonconforming Patient (AMAB)</th>
<th>Pedigree Symbols for Gender Nonconforming Patient (AFAB)</th>
</tr>
</thead>
<tbody>
<tr>
<td>A.</td>
<td>B.</td>
</tr>
<tr>
<td>19.59%</td>
<td>19.18%</td>
</tr>
<tr>
<td>23.69%</td>
<td>21.92%</td>
</tr>
<tr>
<td>49.56%</td>
<td>47.49%</td>
</tr>
</tbody>
</table>

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 genotypic sex 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 actually performing one, similar to the responses that XX or XY would not be appropriate notations for transgender patients.

Participants 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.”

Another participant responded in complete uncertainty, welcoming a new choice all together:

“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”

**Confidence in ability to address psychosocial concerns**

In each of the four scenarios involving a patient identifying as either transgender or GNC comes for genetic counseling, participants were asked “Please rate your level of confidence in assessing the psychosocial needs of your transgender male/transgender female/gender nonconforming patient”. In the scenarios involving a transgender male or female patient, the majority of participants (70.5%) responded that they feel “somewhat confident” in their abilities to address their patient’s psychosocial needs (n = 447). Ten percent felt “very confident and 19.5% felt “not at all confident”. These numbers are not significantly different between addressing the needs of a transgender man and a transgender woman (Figure 3).

When asked the same question in scenarios involving a patient identifying as GNC, patients reported significantly (p<.001) feeling less confident in their abilities to address their patient’s psychosocial concerns in comparison with how they felt about their abilities to counsel a patient identifying as transgender (n = 438). A smaller majority (58.6%) felt somewhat confident in their ability to do this while more (37.5%) responded that they were “not at all confident” in comparison with the responses regarding transgender patients. Only 6.5% reported
feeling “very confident”. Again, these numbers are not significantly different between addressing the needs of a patient who is GNC and was assigned male at birth or female at birth.

**Figure 3: Participants’ confidence in addressing their patients’ psychosocial needs**

Overall, most participants responded that they felt somewhat confident or not at all confident in their abilities to address the concerns of transgender and GNC patients, with only 9.8% feeling very confident. Factors that significantly (p<.002) affected confidence included having had education on the subject, having had a patient who disclosed that they identify as transgender or GNC, knowing someone personally who identifies as transgender or GNC, and having worked in an LGBT clinic. All of these experiences positively correlated with a higher confidence. On the other hand, age (p = .844) and years of patient-facing care (p = .921) showed a slight negative correlation, though this did not reach significance.
Participants were asked what education they have received regarding transgender and GNC healthcare and were given 6 options, of which they could choose multiple (n = 437). 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”. Four hundred and thirty-seven participants responded and 81.5% reported having sought education on the topic, with attending lectures and seeking resources on the internet being the most common choice, followed by published literature and reaching out to colleagues. The remaining 18.5% reported having not had specific education on transgender and GNC healthcare (Figure 4A).

Participants were then asked what educational resources they would like to have regarding transgender and GNC healthcare and were again given 6 options, of which they could choose multiple (n = 437). The options included “online learning modules or webinars”, “lectures”, “workshops”, “more research”, “books or pamphlets”, and “nothing, the education is sufficient”. Six participants felt the “education was sufficient” while the overwhelming majority reported a desire for more education, with “online learning modules or webinars” receiving the most responses Figure 4B).
Participants were asked to assess the risk to a future pregnancy for two transgender patients with a family history of Duchenne Muscular Dystrophy (DMD) (n = 437). The first patient identified as a transgender man, is a known carrier for DMD, and is currently 20 weeks pregnant with a male fetus determined by ultrasound. The overwhelming majority of participants (97.5%) responded correctly that he would have a 50% chance of the fetus being affected with DMD. The second patient identified as a transgender woman who wished to contribute to a pregnancy using banked sperm. Her brother passed away from DMD and their mother is a confirmed carrier. Again, the majority of participants (94.7%) answered correctly that her chance of having an affected male fetus would be closest to 0%.
Discussion

This study served to assess current perspectives 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 experiences with these communities. The education participants have received on transgender and GNC healthcare varied; however, they overwhelmingly 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 impacted by their specialty. Overwhelmingly, genetic counselors only felt somewhat confident in their ability to address the psychosocial concerns of their transgender and GNC patients, which was dependent on if they had received education on the topic and on whether they had personal and/or personal experience with these communities.

Many counselors felt that they may not be as prepared to address potential psychosocial concerns of transgender and GNC patients as they would like. The majority of counselors reported feeling somewhat confident or not at all confident with addressing psychosocial concerns, demonstrating a lot of room for development of these skills. Counselors that did report feeling very confident also reported receiving more education on transgender and GNC healthcare and having more professional and personal experience with transgender and GNC patients. Additionally, the level of confidence counselors reported dropped for GNC patients in comparison to what they assessed themselves as for transgender patients. As counselors get further away from the traditional binary, their understanding of their patients’ experiences starts
to diminish and they feel less confident in their abilities to appropriately address them. Further research into the factors that influence such confidence or lack thereof may provide additional insight on ways to increase counselor competency.

One would expect a greater length of time spent working in the field would yield more confidence in the ability to counsel; however, the opposite was found to be the case when addressing psychosocial concerns in transgender and GNC patients. There was a slight negative correlation between confidence and age as well as confidence and years of experience in their practice area. There could be a number of factors contributing the negative correlation. For one, it may be that genetic counselors who are younger and/or newer to the field are generally more comfortable with the idea of gender fluidity and gender identity outside of the binary. If more education is being provided in training programs, then it would follow that recent graduates would be more confident in addressing these psychosocial concerns. It could also be that less experienced professionals overestimate their ability to completely provide care when asked to gauge their comfort level in a hypothetical scenario. Psychosocial care is complex so it only follows that many factors would impact one’s confidence in their ability to be successful in addressing it in a counseling session.

Within the need for more education, counselors also reported wanting more guidance with regard for pedigree nomenclature for their transgender and GNC individuals. Genetic counselors were generally split between two or three symbols that they felt could be appropriately used. The two most favored symbols for transgender patients reflected the recommendations set forth by NSGC and NCCN. A third one similarly recognized gender identity as the primary symbol, but included more descriptive information underneath. When it came to selecting a symbol for GNC patients, the majority felt that the one that went hand-in-hand with what is 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 to 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.

More important than the actual 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 was particularly important and was the second most-common theme among free responses, speaking to the client-centered approach of genetic counseling. A review of past sociological and therapeutic studies highlighted that acknowledgement and affirmation not only of who they are as they define themselves but also of their struggles has a positive effect on the overall wellbeing of the transgender individual (Connolly, 2005). In their practice of non-directive and patient-focused counseling, genetic counselors are taught to affirm their patients own feelings towards genetic testing and the experiences that led them to feeling that way (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 as they live it.
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 genetic sex. 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 in that footnote unless it has actually been performed. 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. 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 would be. A potential contributing factor to this is the fact that only 20% 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. Another point of confusion 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.”

One thing genetic counselors agreed on in this survey was the desire for more education on transgender and GNC healthcare. Most of the participants were able to accurately assess risk to future children if a transgender man or transgender woman with a family history of DMD were
to be pregnant or donate banked sperm. Even so, some participants did get the questions wrong which could be an indicator of lack of familiarity with terminology. This is one of many circumstances where a counselor could be seeing a transgender patient and they would need to navigate risk assessment when genetic sex and gender identity both play a large part. Our findings demonstrated a positive correlation between having been educated on transgender and GNC healthcare and being confident in one’s abilities to properly counsel a patient. In a study conducted in 2011 assessing the National Transgender Discrimination Survey, at least 50% of respondents stated that they had to educate their providers at the time of being seen in order for them to provide appropriate care (Grant et al, 2011). This takes away from the time actually being able to treat the patient and further marginalizes them. Genetic counselors widely supported more education through readily accessible formats such as online webinars or lectures, workshops, and new research, which is encouraging and 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 psychosocial concerns and use of gender inclusive language. Some participants concluded in their final thoughts in the survey that they want more training and clarity on not offending their patient with their own biases and shortcomings:

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

Others had personal experience with patients who identified as transgender or GNC that highlighted their insecurities of how much they know about the community:

“I just recently had a patient who was a transgender male. ... I had to talk to him about his risk to develop both of those types of cancers based on his mother's genetic testing results. I was totally ill prepared to discuss this with him. I was just so nervous I was going to offend him. And I wanted to respect that he identifies as male, but at the same time, I had to discuss the fact that since he still had female organs, he was at a high risk to develop cancer. I wish I had been able to more comfortably discuss with him his plans for his transition and how these cancer risks
might play a role in his plans. But I just felt very clumsy--we need to hear more from transgender patients about how we should be talking to them about things like this.”

Workshops at both national and regional conferences could address some of these problems. Oftentimes people aren’t 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’ allyship with them. In interviewing transgender and GNC patients who had been to the emergency room, many reported their providers not knowing the term transgender and insensitivity towards their lived experiences (Samuels et al, 2017). In contrast, physicians working in the emergency department reported feeling comfortable asking about personal pronouns and serving transgender and GNC patients despite the majority not having had any education on transgender and GNC healthcare (Chisolm-Straker et al, 2017). These studies exemplify how a lack of self-awareness in healthcare professionals can be detrimental to the patient experience. If genetic counselors were able to ask these questions about their own preparedness and receive guidance from those with more experience, they can ensure that they would come into those encounters with a higher level of understanding.

**Limitations**

One of the limitations of the study was that it was only made available to those 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.

**Future directions**

Counselors are eager for guidance and consistency in how to best address their transgender and GNC patients, which can come from the professional organizations that represent them. Both
NSGC and NCCN can collaborate amongst each other, genetic counselors, and the transgender and GNC communities in order to provide consistent recommendations for pedigree symbols and provide examples of all-inclusive intake forms.

Many genetic counselors in this study reported that they want to be able to provide the best possible care for transgender and GNC patients, but express a desire for more education. Many welcomed the list of terms offered by Fenway health (through a link embedded within the survey instrument) expressed gratitude in having a place to go back to in order to check pronouns and ensure they were using the correct terminology. Healthcare systems, regional, and national organizations that support genetic counselors, such as NSGC and NCCN already provide guidance on many issues surrounding diversity, equity, and inclusion in healthcare and these forums can provide more practical training specific to competency in serving transgender and GNC clients.

Genetic counselors can continue to engage with the transgender and GNC communities on the local and national level and promote more research on health disparities. A general lack of research is apparent throughout many professions within healthcare in addition to genetic counseling. Researching what it is that makes professionals feel unconfident in their abilities to counsel or treat their patients and what those patients want out of these interactions will help many others in healthcare understand their own limitations and give a starting point to build upon them. Education and research can serve as a platform to standardize language, family history intake tools, and gender identity competency.
Conclusions

Over 400 genetic counselors and trainees participated in the survey and the responses illuminated the variability in current practices in pedigree nomenclature as well as self-reported confidence in understanding and addressing psychosocial concerns of individuals who identify as transgender or GNC within genetic counseling. 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 allyship 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 community is divided on appropriate nomenclature that would be most easily interpreted and representative of their transgender or GNC patient on a pedigree, and such variability within current practice could hinder progress towards providing culturally competent care. Confusion and uncertainty on the interpersonal level is likely to hinder client-centered care, thus working towards consistency and competency within the profession will contribute to promoting the best possible care for transgender and GNC individuals seeking genetic counseling services.
Resources


Appendix A: NSGC Recruitment Notice

Dear NSGC Member,

My name is Liz Sheehan and I am a second-year student at Brandeis University’s Genetic Counseling program. I am inviting you to participate in a survey assessing current practices regarding pedigree symbols for transgender and gender non-conforming patients. A second aim of this study is to understand genetic counselor and trainees’ self-reported confidence level in assessing the psychosocial needs of the transgender and gender non-conforming patients, desire for further education, and preferences for educational format.

Study Information:

- All genetics counselors and current students in a Genetic Counseling Training Program are eligible to participate.
- The online survey will take ~10-15 minutes to complete.
- This survey is anonymous. Participation in this study is entirely voluntary and you may exit the survey at any time.
- To thank you for your time, you will have the option of providing your contact information in an unlinked survey to enter a raffle for one of three $50 Amazon Gift Cards. This information will not be linked to your responses.

If you have any questions about this research project, please feel free to contact me at esheehan@brandeis.edu or my advisor, Gayun Chan-Smutko, at gchansmutko@brandeis.edu

Please click here to access the survey:

Thank you for your time and consideration.

Sincerely,

Liz Sheehan
MS Genetic Counseling Candidate
Brandeis University
Appendix B: Survey Instrument

The purpose of this study is to assess current opinions on preferred pedigree nomenclature for representing transgender and gender nonconforming individuals. This survey will assess genetics professionals’ confidence in counseling transgender and gender nonconforming individuals, as well as identify preferences for further education on healthcare for this population.

Useful definitions for taking this survey:

**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 nonconforming**: 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.

**Gender affirmation**: Many people view their coming out as an affirmation of the gender identity they have always had, rather than a transition from one gender identity to another. They may prefer to call themselves “affirmed females” (or just “females”) or “affirmed males” (or just “males”) rather than “transgender” or “transsexuals” because the “trans” prefix suggests they have changed, rather than accepted, their true gender identity. This is consistent with the concept that people do not need to have any surgery in order to affirm their gender.
1. Have you ever worked with a clinic catering specifically to those patients who identify as LGBTQ (Lesbian, Gay, Bisexual, Transgender, Queer)?
   a. Yes
   b. No
2. Have you ever had a patient disclose that they identify as transgender or gender nonconforming?
   a. Yes
   b. No
   c. I’m not sure
3. Do you personally know someone in your life (not a patient) who identifies as transgender or gender nonconforming?
   a. Yes
   b. No
4. Are you aware of any nationally standardized pedigree symbols that represent transgender and gender nonconforming patients?
   a. Yes
   b. No
5. If yes, please cite or provide a description of the resource
6. Please respond to the questions following each scenario
   a. A patient comes into your clinic who identifies as female. In taking her medical history, the patient discloses that her sex assigned at birth was male (transgender female)
      i. Please rate your level of confidence in assessing the psychosocial needs of your transgender female patient.
         1. Very confident
         2. Somewhat confident
         3. Not at all confident
      ii. Which of these pedigree symbols most accurately describes your patient?
         1.
         2.
         3.
b. A patient comes into your clinic who identifies as male. In taking his medical history, the patient discloses that his sex assigned at birth was female (transgender male)
   i. Please rate your level of confidence in assessing the psychosocial needs of your transgender male patient.
      1. Very confident
      2. Somewhat confident
      3. Not at all confident
   ii. Which of these pedigree symbols most accurately describes your patient?
iii. Please explain why you believe that to be the best choice for this scenario.

iv.

c. A patient comes into your clinic who identifies as gender nonconforming (GNC), identifying as neither male nor female. In taking their medical history, you learn that their sex assigned at birth was male.

i. Please rate your level of confidence in assessing the psychosocial needs of your gender non-conforming patient
   1. Very confident
   2. Somewhat confident
   3. Not at all confident

ii. Which of these pedigree symbols most accurately describes your patient?

   1.

   2.

   3.

   4. XY

   5. Other - please describe with words
iii. Please explain why you believe that to be the best choice for this scenario.

d. A patient comes into your clinic who identifies as gender nonconforming (GNC), identifying as neither male nor female. In taking their medical history, you learn that their sex assigned at birth was female.

i. Please rate your level of confidence in assessing the psychosocial needs of your gender nonconforming patient.

1. Very confident
2. Somewhat confident
3. Not at all confident

ii. Which of these pedigree symbols most accurately describes your patient?

1. 

2. 

3. GNC, XX

4. 

5. Other - please describe with words

iii. Please explain why you believe that to be the best choice for this scenario.

7. You have a patient who is 20 weeks pregnant referred to you for a family history of Duchenne Muscular Dystrophy (DMD), which is an X-Linked condition. The patient identifies as a transgender male (female sex assigned at birth). The patient is a known carrier of DMD. The sex of the fetus is determined to be male by ultrasound. What is the risk for the fetus to be affected with DMD?

a. 0%
b. 25%
c. 50%
d. 100%
8. You have a 30 year old patient who was referred to you for a family history of Duchenne Muscular Dystrophy (DMD), which is an X-Linked condition, and would like to undergo preconception counseling. The patient identifies as a transgender woman (male sex assigned at birth). Prior to transitioning, the patient stored their sperm in hopes of having a biological child with their partner. In taking the family history, you find out the patient’s brother died of DMD at 22 years old and their mother is a known carrier. What is the risk to have a child affected with DMD?
   a. 0%
   b. 25%
   c. 50%
   d. 100%

9. What kind of education have you had on transgender or gender nonconforming healthcare? (Select all that apply)
   a. I have attended or listened to lectures
   b. I have attended a workshop
   c. I have read scholarly articles or books on the topic
   d. I have sought out my own sources on the internet
   e. I have reached out to colleagues who may be more knowledgeable
   f. I have not had any specific education on the topic

10. What kind of educational resources would you like to see more of regarding transgender and gender non-conforming healthcare? (Select all that apply)
    a. Lectures
    b. Workshops
    c. On-line learning modules or webinars
    d. Books or pamphlets
    e. More research
    f. Nothing, the education is sufficient

11. What gender do you identify as?
    a. Cisgender Female
    b. Cisgender Male
    c. Transgender Female
    d. Transgender Male
    e. Gender Non-Binary
    f. I do not wish to disclose

12. How old are you?
    a. <22
    b. 22-35
    c. 36-49
    d. 50-63
    e. 64-70
    f. >70

13. Do you currently provide patient-facing care as a genetic counselor?
    a. Yes, currently
    b. Yes, in the past but not now
c. Yes, as a student in Genetic Counseling
  d. No

14. How many years have you provided patient-facing care?
   a. 1-5
   b. 6-10
   c. 11-15
   d. 16-20
   e. 20+
   f. I’m currently a student

15. In what field of Genetics do you practice?
   a. Oncology
   b. Pediatrics
   c. Prenatal
   d. Adult/General
   e. Other specialty clinic
   f. Lab/Research/other

16. How many years have you been practicing in the field you currently work?
   a. 1-5
   b. 6-10
   c. 10-15
   d. 15-20
   e. 20+
   f. I’m currently a student

17. How many new patients do you see a week, on average?
   a. 1-4
   b. 5-8
   c. 9-12
   d. 13-16
   e. More than 16

18. What percent of your time do you spend on clinical activities?

19. Do you have any other comments regarding these topics?