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Title: Trans-Inclusive Genetic Counseling Services: Recommendations from Members of the Transgender and Non-Binary Community

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Abstract

The term transgender is used to describe individuals whose gender identity does not align with their sex assigned at birth. The term transgender can include individuals who identify as male, as female, as both of the traditional concepts of masculine and feminine gender, or neither masculine or feminine. The latter two gender identities may also be known as non-binary or gender non-conforming identities. Transgender individuals may attend genetic counseling for a variety of reasons, but current pedigree nomenclature does not adequately represent both sex assigned at birth (which is important for determining risk for certain conditions) and gender (which is important for providing trans-inclusive care). We conducted an interpretive description (qualitative, interview based) study with individuals from the transgender community to gather insight on pedigree nomenclature and more broadly, how to provide safe and effective genetic counseling for transgender individuals. We conducted semi-structured telephone interviews with individuals who identified as transgender or gender non-binary, transcribed them verbatim, and checked them for accuracy before coding and inductively identifying themes. Among our 8 participants, 5 identified as trans-masculine, 2 as trans-feminine, 5 as non-binary/gender non-conforming (some participants had more than one gender identity). From the interviews, we identified a single key, over-arching theme: participants' felt it is the genetic counselor's responsibility to create safety and provide clarity about the clinical importance of both sex assigned at birth and gender identity for trans patients. Two specific strategies that counselors could use to achieve this safety and clarity were discussed extensively: 1) validating gender identity and 2) using inclusive and well-defined pedigree symbols that denote both sex assigned at birth and gender identity. Our data have important practice implications in terms of the importance of validating gender identity and using respectful pedigree symbols.

Key Words: transgender, non-binary, gender non-conforming, pedigree symbols, pedigree nomenclature; family history; genetic counseling, trans-inclusive, trans health; sex; gender; interpretive description; LGBTQ+; risk assessment; cultural competence; diversity; underrepresented populations

Introduction

Despite frequent conflation in medicine, the terms “sex” and “gender” are not interchangeable. Sex refers to biological attributes and can be defined by variables such as body morphology (e.g. the appearance of genitalia or secondary sex characteristics), chromosomes, or hormones. Gender is a social and cultural construct that ascribes different roles, characteristics, and values in relation to the sex assigned at birth. The term “gender identity” is used to describe an individual’s personal concept of their gender, such as identifying as male, female, both male and female, neither male or female, or something else (Davidson, 2015). The term “transgender” can be used to describe individuals whose gender identity does not align with their sex assigned at birth (Griffin, 2017). The term transgender can sometimes include individuals who do not identify with either male or female gender identities; these individuals may use terms like “non-binary” or “gender non-conforming” to reflect that they do not identify with the binary nature of male and female gender identities (GLAAD, 2011). During an individual’s transition process, transgender and non-binary individuals may undergo medical transitions such as gender-affirming surgery and/or hormones, a social transition consisting of pronoun/name change and/or a change to their gender expression (e.g., clothing, hair style, etc.), both medical and social transition, or neither a medical or social transition. For a glossary of terms used in this paper, see Table 1.

<<Insert Table 1 about here>>

Outside of accessing healthcare for reasons directly related to their trans identity, transgender individuals will access healthcare services – including genetic counseling – for a variety of reasons. However, data shows that transgender individuals access healthcare services less than others, due to feeling unsafe and experiences of stigmatization of their gender identity

(Clark, Veale, Greyson, & Saewyc, 2018; Giblon & Bauer, 2017; Merkel, 2017; Poteat, German, & Kerrigan, 2013; Puckett, Cleary, Rossman, Mustanski, & Newcomb, 2018). For example, experiences with misgendering – when an individual is referenced or spoken to with pronouns or a name that does not align with their gender identity – can lead to increased levels of depression, reduced self-esteem, and feelings of stigmatization (McLemore, 2015; McLemore, 2018). When experienced in a healthcare setting this can in turn can result in decreased future access to healthcare services, which can compound mental and/or physical health problems (Davidson, 2015; White Hughto, Reisner, & Pachankis, 2015).

The genetic counseling encounter aims to help patients to “understand and adapt to the medical, psychological and familial implications of genetic contributions to disease” (Resta et al, 2006). One of the key components of the genetic counseling session is the construction of an accurate pedigree - a visual representation of an individual’s family history. Collecting a family history in the form of a pedigree can be useful in building rapport, evaluating the patient’s needs for the session, to understanding the inheritance patterns of disease, providing accurate risk assessment to patients and their family members, and understanding familial relationships. To construct a pedigree, the counselor asks the patient questions about their own and their family members’ health history. Questions are typically targeted towards the reason for referral – e.g. in the context of a genetic counseling session about cancer, questions will routinely be asked about whether cancers after an initial diagnosis were additional primary cancers or metastases, age at first diagnosis, family history of other cancers, etc. Individuals are represented on pedigrees as symbols following a standardized set of nomenclature for the generation of pedigrees by the National Society of Genetic Counselors (NSGC) (Bennett, French, Resta, & Doyle, 2008). These symbols are standardized to allow for correct interpretation and risk assessment and avoid

misinterpretation of symbols by future healthcare providers who may use the pedigree (Bennett, 2008). Circles and squares are the most frequently used symbols on pedigrees and are typically used to refer to cisgender women and cisgender men, respectively. Other standardized symbols include the triangle (e.g., for spontaneous or therapeutic abortions) and the diamond (uses discussed further below).

The current recommendations of the NSGC regarding standardized nomenclature indicates that the diamond can be used as the symbol to represent transgender individuals. However, the diamond can also be used in situations where “it is not clinically relevant to assign gender”, “gender is not specified”, and for congenital disorders of sex development (Bennett, 2008). The diamond does not allow for the representation of either the individual’s gender identity or their sex assigned at birth, and one cannot distinguish between a trans man, a trans woman, or a non-binary individual. An alternative suggestion provided in the guidelines is to represent the individual with a symbol representing their “phenotypic gender” with the karyotype noted below if it is known and available (Bennett et al., 2008). However, given that karyotypes are not necessarily routinely available this solution is pragmatically problematic. Further, the meaning of the term “phenotypic gender” in this context is unclear. It could refer to how individuals choose to present themselves to the world (i.e. their gender expression, which may be reflected in choice of clothing worn to the appointment). Alternatively, if the term “gender” has been used as a synonym for “sex” it could refer to body morphology and/or anatomy. Either way, there is no clear guidance around how to capture both sex and gender for transgender individuals.

The National Comprehensive Cancer Network (NCCN), which provides guidelines on the most up-to-date management and treatment for cancer patients (typically cisgender patients), included two specific symbols for “transsexual” individuals in a 2017 updated guideline for

colorectal cancer. The symbols are a combination of the square and circle with one inside the other, where gender identity is represented as the outer symbol and sex assigned at birth is the inner symbol (Provenzale et al., 2016). However, it is unclear what sources were used to derive these symbols (though the authors cite a previous paper in relation to the origin of the symbols, it does not mention transgender or transsexual individuals at all). As well, not everyone who is transgender identifies with the term “transsexual”; indeed, it is regarded as an offensive and outdated term by some members of the transgender community (GLAAD, 2011). The two symbols proposed by the NCCN are an improvement over the diamond in that they represent both gender identity and sex assigned at birth, but they do not accommodate those who identify as non-binary and rely on problematic terminology.

It is not clear if transgender individuals were consulted in the creation of any of the symbols that have been proposed to symbolize this population in pedigrees. However, such consultation is critical – members of socially marginalized groups have previously shared the importance of their input and experiences being heard and respected in creating healthcare plans and procedures that service members of their community (Snow, Tweedie, & Pederson, 2018). The inclusion of members of these marginalized groups, such as the LGBTQ+ population, can lead to a more inclusive delivery of healthcare services related to the specific health needs and wellbeing goals of that community (Mulé et al., 2009). As such, the representation and inclusion of transgender and non-binary individuals in research regarding the symbology used to represent them on pedigrees is an essential part of developing a trans-inclusive service.

Given the challenges associated with the pedigree symbols currently suggested for transgender individuals, there is the potential for genetic counseling to feel unsafe for transgender and non-binary individuals. For example, a patient who is transgender may feel

invalidated or less trusting of a genetic counsellor who hesitates and is unsure what symbol to use to represent a transgender individual on a pedigree, when cisgender patients are quickly symbolized by a square or circle without hesitation. Thus, we sought to engage with members of the transgender community to explore their perspectives on ensuring trans-inclusivity practices in genetic counseling to provide safe and validating care. The aims of the study were to gather feedback and recommendations from individuals from the transgender community on appropriate practice as it pertains to discussing sex and gender related issues in genetic counseling practice, and to gather suggestions for appropriate and respectful pedigree symbols to represent transgender individuals.

Methods

We conducted a qualitative, interpretive description study in which we conducted telephone interviews with members of the transgender community to gain insight from their perspective on safe, trans-inclusive genetic counseling services. The interviewer was the first author, who was a genetic counselling student at the time of the interviews. The authors are all white genetic counselors who were assigned female sex at birth, and express a gender that aligns with their birth sex. This study was approved by the Children's and Women's Research Ethics Board at the University of British Columbia in Vancouver, Canada (H18-01386).

Participants and Procedures

Participants were recruited for this study through advertisements on social media. After potential participants indicated their interest (via email to the first author), eligibility was

confirmed (self-identified as transgender, fluent in English, >18 years old), and consent was obtained.

Participants were provided with a four-minute YouTube video from the NSGC entitled “What Is A Genetic Counselor?”¹ and a one-page document that explained the current utility of pedigree symbols in genetic counseling, including the square, circle, diamond, and triangle (see supplemental material). After participants had reviewed these materials, semi-structured telephone interviews were conducted; telephone interviews allowed individuals from a wider geographic area to participate and reduced the possibility of visual judgement that may occur in in-person interviews. The interviews explored participants’ experiences with healthcare providers as it pertained to their gender identity and any experiences with genetic counseling (if applicable; see supplemental material). The participants were asked for their feedback on the current use of pedigree symbols and they gave suggestions for appropriate symbols to use for transgender and non-binary individuals. In addition, two scenarios where the distinction between sex assigned at birth and gender identity were crucial components of effective and accurate genetic counseling were presented to the participants for their feedback (see Figures 1 and 2).

<<Insert Figures 1 and 2 about here>>

Participants also provided information about their demographics, gender identity and pronouns with questions adapted from a recommendations guideline for including trans-inclusive questions on population surveys (Bauer, Braimoh, Scheim, & Dharma, 2017). The interviews were transcribed verbatim and checked for accuracy. Participants received a \$15 CAD Amazon gift card for their participation upon completion of the interview.

¹<https://www.youtube.com/watch?v=GDjLazXGV0s>

Data Analysis

Originating in a nursing context, interpretive description is a qualitative research approach that studies a phenomenon through thematic analysis of a small group of participants, examining their experiences and perspectives on that phenomenon (Thorne, S., Kirkham, & MacDonald-Emes, 1997; Thorne, Sally, Kirkham, & O'Flynn-Magee, 2004). Studying individual cases allows insight to the meaning and reasons behind participants' experiences with the phenomenon, allowing for the generation of common themes (Thorne et al., 1997; Thorne et al., 2004). The common themes derived from the smaller cohort can be used in the formation of a clinical understanding of the phenomenon that can be, in turn, applied to other individual cases (Thorne et al., 1997; Thorne et al., 2004). Specifically, analysis starts by immersion in the data as it is gathered, and coding at the level of smallest units – e.g. sentences. Memos and linkages are generated iteratively, which allows the researcher to synthesise and contextualize ideas, and theorize about their relationships within and between participants (Thorne et al., 1997; Thorne et al., 2004).

Throughout data collection, all authors discussed concepts and ideas as they emerged from the interviews. Following completion of interviews, three transcripts were coded by two coders (the first and second authors), and discussed and checked for consistency and agreement, before the first author applied the coding framework to the remaining transcripts for the creation of themes. Recruitment was terminated when saturation of main themes (i.e. when no new concepts or ideas were identified from interviews with additional participants) was achieved. Upon completion of the interviews, preliminary themes were circulated to participants to solicit feedback on whether they accurately captured their perspectives. After reviewing the feedback

from the participants, findings were restructured to condense three separate themes into one central theme with two sub-components, or strategies.

Results

We interviewed eight transgender individuals, some of whom had sought and received hormones and/or surgeries as a part of their transition and some who were not pursuing surgical or hormonal interventions. All participants were white and had some level of university education at the time of the interview (demographic details provided in Table 2 and Table 3). Interviews ranged from 32 to 60 minutes in length.

<<Insert Tables 2 and 3 about here>>

A single key overarching theme was identified: that it is the responsibility of the genetic counselor to create safety and provide clarity regarding the importance of both sex and gender to the genetic counseling appointment. Two specific strategies that counselors could use to achieve this safety and clarity were discussed extensively: 1) validating gender identity and 2) using inclusive and well-defined pedigree symbols that denote both sex and gender. All of these concepts were endorsed by participants through our member checking process.

It is the responsibility of the Genetic Counselor to create safety and provide clarity

Participants clearly articulated that it is the role of the genetic counselor to provide a safe environment for their transgender patients, and to provide as much clarity as possible about the importance of both sex and gender in terms of the service they are providing. Participants felt that part of the creation of a safe environment and providing clarity for patients involves the

genetic counselor to be able to think and talk about the distinction between and clinical importance of sex and gender, and to use terms appropriately. For example:

There's a big difference in being like, 'You have an XY chromosome and so that puts you at an elevated risk,' versus saying, 'You are a man and that puts you at an elevated risk.' It's just a change in language ... There's nothing inherently transphobic or anything about saying, 'XY chromosomes have this impact' ...in the same way, 'If you have a cervix you can get cervical cancer' ...as opposed to saying 'Women get cervical cancer.'(Taylor, non-binary)

I do think that the genetic counselor should be able to talk about [gender identity vs. sex assigned at birth] with the patient just because, really, like obviously, the patient should be knowledgeable about their own health, but it's not really like their job to know that [the implications of gender identity and sex assigned at birth] is important and that is something that [the genetic counselor] need[s] to know. (Nicholas, transmasculine and non-binary)

Other participants noted that as genetic counselors are providing information that has implications for the extended family, ascertaining the correct sex assigned at birth and gender identity for all individuals in the pedigree is important. Establishing normative clinical practice of routinely asking all patients about sex and gender also establishes the clinical environment as a safe space for transgender individuals, because the questions are not being applied only to individuals who a genetic counselor believes is transgender. This point is illustrated by the following quotes from two of the participants:

I think that the [genetic counselor] probably should not be assuming that every person within a family unit is this gender and should probably make some inquiries as to, you know, "Hey you said you had two sisters, are they both – or were they both assigned female at birth?" (Mara, transfeminine)

If you don't ask everyone about their pronouns and you only ask people about their pronouns when you suspect they're transgender, all you're doing is proving that you can spot a transgender person. You know what I mean? So making it kind of like standard practice with everyone. (Taylor, non-binary)

Strategy One: Validation of Gender Identity is Key

Participants clearly identified that one of the ways in which healthcare professionals could fulfill their responsibility to create a safe environment for trans patients was by validating their patient's gender identity – this was raised by all participants to varying degrees. Some expressed that they had been made to feel invalidated or unsupported by healthcare providers in the past in this regard, and that this had influenced both how they felt about accessing healthcare since, and how they engage with healthcare professionals. Aaron, an individual identifying as transmasculine and gender non-conforming shared, “I tend to avoid doctors as much as possible. But I have this situation where I had to go see a family doctor for like a physical for my top surgery a few years back and my family doctor refused to see me about it.”

The importance of a safe environment in healthcare for transgender individuals is further illustrated in the following two quotes:

I avoided going to the doctor until I found one was affirming ... basically I would go to the doctor if I had no other options or an emergency for a long time, and then when I heard that there were affirming doctors in the area that is when I actually set myself up with a primary care provider and started getting routine medical care. (Mara, transfeminine)

I think that part of it, like, I would, if I were choosing a new doctor, for example, if I moved somewhere, I would want to make sure they had worked with trans people before solely so I wouldn't have to explain myself, because it gets exhausting. So that's probably part of it is going into it knowing they already know the terms, and they already know what it's like to work with a trans person. So I don't have to be the first one and I don't have to sit there defining all the things to them because that's always the worst. (Parker, transmasculine and non-binary)

Participants reported that one of the key factors in feeling safe and respected as a transgender individual in a healthcare environment, was to have their gender identity be treated as valid and not as something strange or unknown. One participant shared a practical example of a respectful and inclusive practice, that can create a safer environment for trans patients from the outset:

Having that space [on forms for pronouns and preferred name] where someone could include something that isn't just on the binary. Things like that that allow them, right off the bat, to realise that okay, at least there's some attempt to be, kind of, inclusive of myself [...] to at least take that chance with them, I guess, that certainly helps. (Edward, transmasculine)

Participants emphasized that mistakes (e.g. use of the wrong pronouns or name) happen, but it is how they are handled that is key. Two participants shared how a brief respectful apology and moving on is the best strategy:

We all make mistakes and we kind of expect people are going to make mistakes and that's okay. I make mistakes all the time myself but, yeah, it's just that acknowledgement and apology, and then to really try to change it. It's very different than someone who just kind of brushes it off and keeps making the mistake. But you can kind of tell when someone's really not making an effort. But as long as there's an effort there, a recognition, kind of a quick apology, that's ... to me I think that's something that seems to go ... well. (Edward, transmasculine)

In regards to getting misgendered, it's way more bothersome if someone makes a big deal out of it. Like, oh my God, I'm so sorry. I'll never do it again. That doesn't really help anyone. It just makes everyone feel awkward, and it makes you feel bad. So it's way easier in any situation, whether it's a healthcare provider or not, to like just apologize and correct yourself and move on.” (Parker, transmasculine and non-binary)

Strategy Two: Pedigree Symbols must be Inclusive and Well-defined

Participants discussed how genetic counselors could fulfill their responsibility to provide clarity of the importance of both sex and gender by using pedigree symbols that are inclusive,

and well defined. With this in mind, participants talked about how the use of the diamond does not feel clear because by itself does not distinguish between trans men, trans women, and non-binary individuals:

Like, there's a difference between someone who's FTM [female to male] and someone who's MTF [male to female]. So I wouldn't really want to use the same symbol for both because, yes, I get it's kind of an umbrella, but there are so many different things that go on underneath it that I don't really think you could put it all down to one thing. (Parker, transmasculine and non-binary)

Participants also talked about how the diamond did not feel validating, given that it can also be used when gender is not clinically relevant, as shared by Edward, a transmasculine participant:

Like why ... because my gender doesn't kind of conform to the normal ... or to the ... what's considered normal male, female, why is it now put in the same category as not important? [...] In terms of what a person could feel, like that kind of invalidation ... almost like a non-important feeling. (Edward, transmasculine)

While some participants felt that pedigree symbols should be used to denote only sex assigned at birth or reproductive ability, the majority of participants felt that it was critical to have both gender identity and sex assigned at birth represented within the pedigree. Nicholas, a transmasculine and non-binary participant, shared his opinion on what kind of symbol he would like to represent himself:

Probably some sort of male symbol that indicated that I was trans, I would definitely prefer that over a symbol indicating that I was female. I'd much rather it would be either like essentially female transition to male, then just represented as just female. (Nicholas, transmasculine and non-binary)

Five options for pedigree symbols were provided by participants in the study (see Figure 3).

<<Insert Figure 3>>

Two of the proposed symbols were excluded by the researchers for pragmatic reasons such as symbol identification could be challenging if shapes were drawn freehand (e.g. the half-circle/square, half diamond option shown in Figure 3), interpretation confounded if shaded in (e.g. the circle/square/diamond with a T in the middle shown in Figure 3). The remaining three proposed symbols were returned to participants for additional feedback. The preferred option for most participants involved the use of a single shape to represent gender identify (masculine=square, feminine=circle, non-binary= diamond) with the annotation “AFAB” or “AMAB” to indicate “assigned female/male at birth”. Participants who chose this option felt that this was clearer and less ambiguous than the other options and they liked that gender identity is more prominently represented than sex assigned at birth. Participants also appreciated how with this symbol, for example, a cis woman and trans woman are both represented with the same symbol, with the sex assigned at birth still being clearly documented with AMAB/AFAB. Participants who did not choose this option as their first choice expressed concern about whether interpretation of these symbols would be consistent.

Discussion

This is the first study of which we are aware to qualitatively engage with members of the transgender community to generate symbol designs that they would feel comfortable with representing their sex assigned at birth and gender identity on pedigrees, and to explore how to make genetic counseling trans-inclusive. We heard clearly from our participants that they feel genetic counselors have a responsibility to create a safe environment and to provide as much clarity as possible around gender and sex and their relevance to the service they are receiving. Broadly, these concepts align well with and are supported by findings of previous work

exploring the genetic counseling experiences of other members of the LGBTQ population (specifically, gay, lesbian, and bisexual individuals) (VandenLangenberg, Leach, LeRoy, & Glessner, 2012).

Creating a safe healthcare environment is critically important component for transgender individuals to feel comfortable seeking medical care (Grant et al., 2011). Some strategies for creating a safe healthcare environment are inclusive posters/pamphlets around the clinic, forms with spaces for pronouns and preferred name, and clinic staff and healthcare provider education on transgender healthcare and social terminology such as pronouns and names (Deutsch, 2016; Grant et al., 2011). Creating this sense of safety can come from providing, as our participants suggested, validation of gender identity. This is a crucial step; our participants discussed how the validation of their gender identity is necessary for them to be able to engage in their healthcare. Indeed, the importance of creating a safe environment for trans and non-binary people cannot be overstated. For example, the Canadian Trans Youth Health Survey showed that higher levels of trust and comfort with a healthcare provider correlated with better self-reported mental and physical health (Clark et al., 2018; Veale et al., 2017). On the other hand, research shows that transgender individuals access healthcare services at lower rates, and that this is related to stigmatization of their gender identity (Clark et al, 2018; Giblon & Bauer, 2017; Merkel, 2017; Poteat et al, 2013; Puckett et al 2018). This decreased access to healthcare services can lead to poorer health outcomes, both physically and mentally (Davidson, 2015). Therefore, it is reassuring that in other previous work, genetic counselors have reported feeling that creating a safe environment and validating gender identity are key components of providing appropriate care for transgender patients (Sheehan, 2018). Practical considerations and recommendations on how to create a safe environment for culturally competent care are illustrated in Table 4.

The genetic counselor's responsibility to provide as much clarity about the importance of both sex and gender as possible is especially important in situations where the accurate delivery of medical information depends on accurate identification of a patient's sex assigned at birth. Sex assigned at birth may or may not align with the embodied gender identity of a patient, and when we simply ask a patient about "brothers and sisters" we do not actually know whether a reported brother was actually assigned the female sex at birth. Therefore, it is crucial that the counselor explicitly establishes sex as assigned at birth (rather than making assumptions) in order to provide accurate medical information. Further, in some instances, decision making may be influenced by sex and gender related issues – for example, decision making around surgeries for transgender individuals who carry pathogenic mutations in *BRCA1* or *BRCA2* may be influenced by both efforts to reduce risk for cancer and to affirm gender as a part of their transition (Sacca, Koeller, Rana, Garber, & Morganstern, 2019).

Three of the options for pedigree symbols suggested by our study participants provide information on both the gender identity and sex assigned at birth of the represented individual, which is an essential component of providing trans-inclusive genetic counseling. Of the three options, the one favoured most strongly by participants in our member checking process was the use of squares/circles to represent masculine/feminine gender identity, and diamonds to represent non-binary individuals, with the annotation "AMAB/AFAB" outside the symbol to indicate "assigned male at birth/assigned female at birth" respectively. While this feels like an appropriate way of indicating transgender and non-binary individuals on pedigrees, it does have more broadly reaching implications for pedigrees in general.

Given that current guidance is unclear if squares and circles currently represent sex or gender in pedigrees, and our data suggest that the shapes should represent gender, and that sex

should be indicated with an annotation, perhaps this same strategy should be applied in general. As an example of a way to clarify the meaning of symbols, shapes could be used to represent gender and perhaps those whose sex aligns with their gender could be indicated with a C (for “cis”) by the symbol, or more simply, the pedigree could include a checkbox indicating that “all individuals for whom sex assigned at birth does not align with their current gender identity are indicated on the pedigree”, which would allow any reader to assume that if a symbol does not include an “AMAB/AFAB” annotation, sex assigned at birth and gender identity align.

Implementing an approach such as this would also be in the spirit of our data, as our participants emphasized the importance of the genetic counselor not making any assumptions about anyone’s sex or gender, and asking all patients questions surrounding sex assigned at birth and gender identity. A list of potential questions during the generation of a pedigree for both transgender and cisgender patients are displayed in Table 4.

<<Insert Table 4 about here>>

The other broader consideration raised by using symbols as described here relates to the use of diamonds in pedigrees for purposes other than indicating non-binary gender identity – i.e. when used with a particular number or an “n” inside it, it can be used to represent a group of male/female/masculine/feminine relatives. We feel that the diamond with a number inside it can still be used for this purpose, and should be readily distinguishable from an individual who identifies as non-binary, with the use of the AMAB/AFAB acronym as a part of the symbol for a non-binary individual indicating that distinction.

Practice Implications

Our data show that to provide a safe environment for transgender patients, it is important for genetic counselors to validate gender identity, and to be clear about the clinical importance of both sex assigned at birth and gender identity. Our work suggests that changes to pedigree nomenclature guidelines may be necessary, and indeed, work is currently ongoing to revise the standard pedigree nomenclature. Further recommendations on how to address the subject of sex assigned at birth and gender identity, as well as suggestions for increasing a trans-inclusive service, are included in Table 4.

Taken together with the findings of other studies where genetic counselors note their uncertainty and anxiety regarding counseling for members of the LGBTQ+ population, to provide a trans inclusive environment further education and training for both genetic counselors and graduate students may be important (Sheehan, 2018; Gamma, Ward, & Gallagher, 2015; Glessner, VandenLangenberg, Veach, & LeRoy, 2012; Zayhowski et al., 2019).

Study Limitations

One of the main limitations is that all the participants in this study were white. Transgender and non-binary individuals of color often experience higher rates of discrimination in many aspects of their life, such as employment, education, and healthcare settings – all of which can negatively impact their physical and mental health (Grant et al., 2011; James et al., 2015). Individuals of color may also have differing views on the concept of gender itself as opposed to the binary concept seen prominently in western culture, such as the two-spirit identity of some Indigenous cultures (Filice, 2015). Interpretive description uses the experiences of individual cases to generate common themes to form an understanding of the phenomenon, and as the experiences of transgender and non-binary individuals of color may be different than the

participants in this study, their opinions and perspectives on genetic counseling and symbol usage may differ.

Our participants were all employed to some degree and were well-educated, with most having achieved at least some university education at the time of the interviews. The majority of the participants identified as transmasculine or non-binary, with only two participants identifying as transfeminine, and most of our participants were young adults in their twenties. Individuals of different socioeconomic statuses, education levels, gender identities, and ages may have provided different perspectives on building a trans-inclusive genetic counseling service.

This study did not include the perspectives of intersex individuals as the aims of this study were specifically focused on transgender and non-binary individuals. There are current recommendations for pedigree symbols for intersex individuals, such as a circle or square with the karyotype denoted below (Bennett, 2008), and feedback on these symbols and the discussion of safe and inclusive genetic counseling services for intersex individuals would require input from members of this community.

Future Research

Future research may include a quantitative study to gather wider feedback and suggestions from individuals from the transgender community about pedigree symbols and appropriate genetic counseling care. Importantly, future research should include transgender and non-binary individuals of color to provide their feedback and perspectives, as well as individuals from different socioeconomic statuses, education levels, ages, and gender identities. Further research into the area of pedigree symbols and inclusive genetic counseling practice for intersex individuals is also recommended.

Other future directions may also include a survey of genetic counselors on the utility of the suggested pedigree symbols, including their effectiveness with transgender and non-binary individuals and the practical application within the counseling session.

Author Contributions

HB: contributed substantially to the design of the work, acquisition of data, analysis and interpretation of the data, drafted the manuscript, approved the final version and agrees to be accountable for all aspects of the work

EM: contributed substantially to the analysis and interpretation of the data, revised the manuscript for important intellectual content, approved the final version and agrees to be accountable for all aspects of the work

JA: contributed substantially to the design of the work, analysis and interpretation of the data, co-drafted the manuscript, approved the final version and agrees to be accountable for all aspects of the work

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Conflict of Interest: HB, EM, and JA declare that they have no conflicts that could bias this work.

Human Studies and Informed Consent: 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 individual participants included in the study. The study was approved by the University of British Columbia Children's and Women's Research Ethics Board (H18-01386).

Animal Studies: No animal studies were carried out by the authors for this article.

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Table 1. Glossary of Terms.

Term	Definition
Cis/cisgender	An individual whose gender identity aligns with the sex they were assigned at birth.
Gender expression	How someone chooses to express their gender identity, i.e. through name, pronouns, clothing choices, hair styles, etc.
Gender identity	An internal sense of one's gender.
Gender non-conforming	Individuals whose gender expression varies from the typical expectations of masculine or feminine gender roles.
LGBTQ+	An acronym standing for Lesbian, Gay, Bisexual, Transgender, Queer/Questioning, Other (referring to other gender identities/sexual orientations). There are many variations of this acronym.
Misgender	Referring to a transgender individual by the incorrect pronouns, name, or gender; this may be done either intentionally or accidentally
Non-binary	A gender identity that does not identify with either the binary male or female gender identities, but rather as something between or outside of these identities.
Trans/Transgender	An individual whose gender identity or gender expression does not align with the sex they were assigned at birth. Transgender individuals can use the terms 'MtF' (male to female), FtoM (female to male), transmasculine, transfeminine, non-binary, gender non-conforming, genderqueer, etc. to describe their gender identity.

Figure 1. An X linked condition. John, the proband, does not mention that his sister Rebecca is transgender, and she is represented as a circle. This scenario focused on the screening and testing recommendations that were provided for John and his brother Robert, but did not touch upon his sister Rebecca who is also at risk for this condition. John was counseled that his sisters are at risk to be carriers and carriers are typically unaffected.

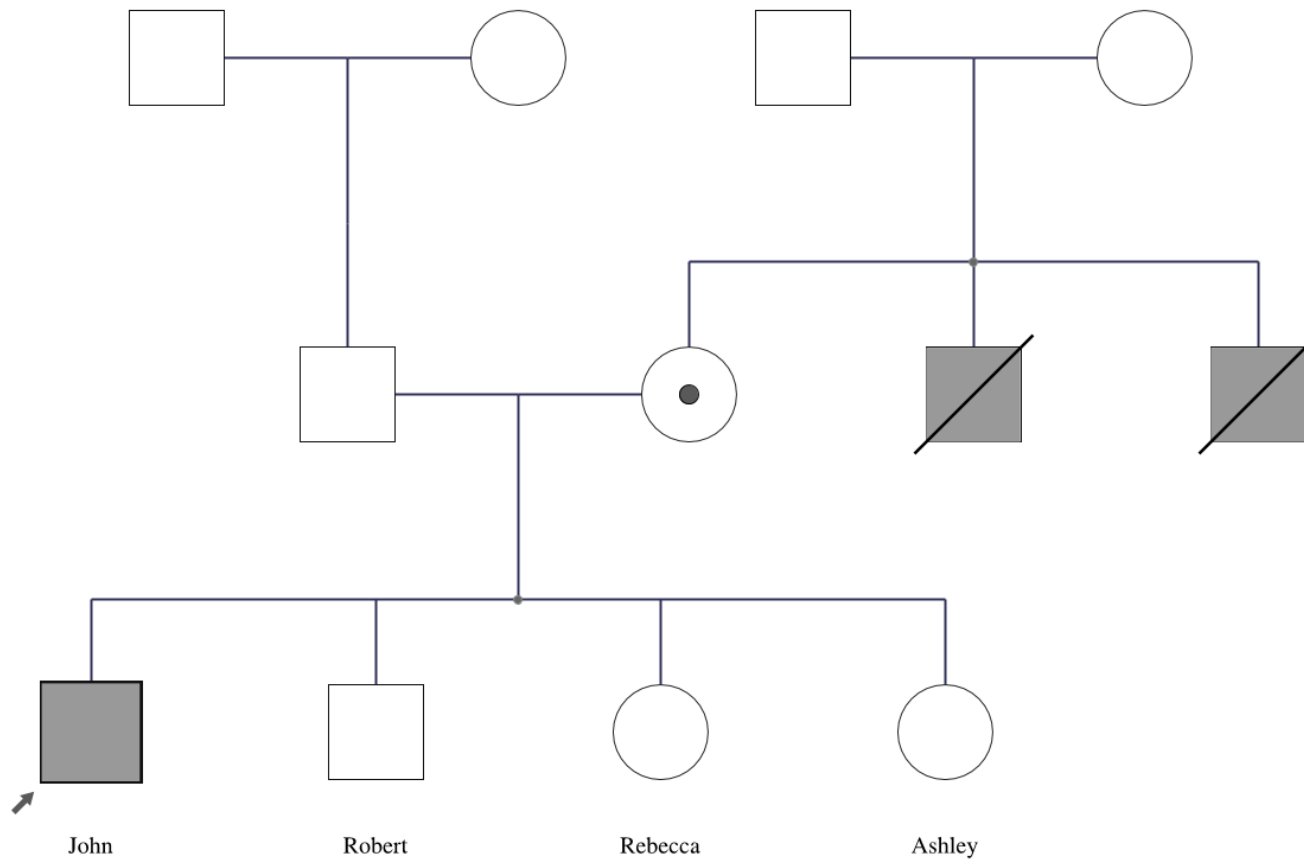


Figure 2. A family with Hereditary Breast and Ovarian Cancer. Paul is transgender and is represented by his “phenotypic gender” of male by the first genetic counselor they see. This scenario focused on a situation where genetic testing in his affected sister Kaitlyn revealed no mutation. As a result, the implications for screening and prophylactic surgeries for Paul based on his family history were not provided accurately by a second genetic counselor due to the misrepresentation of his gender identity on the pedigree.

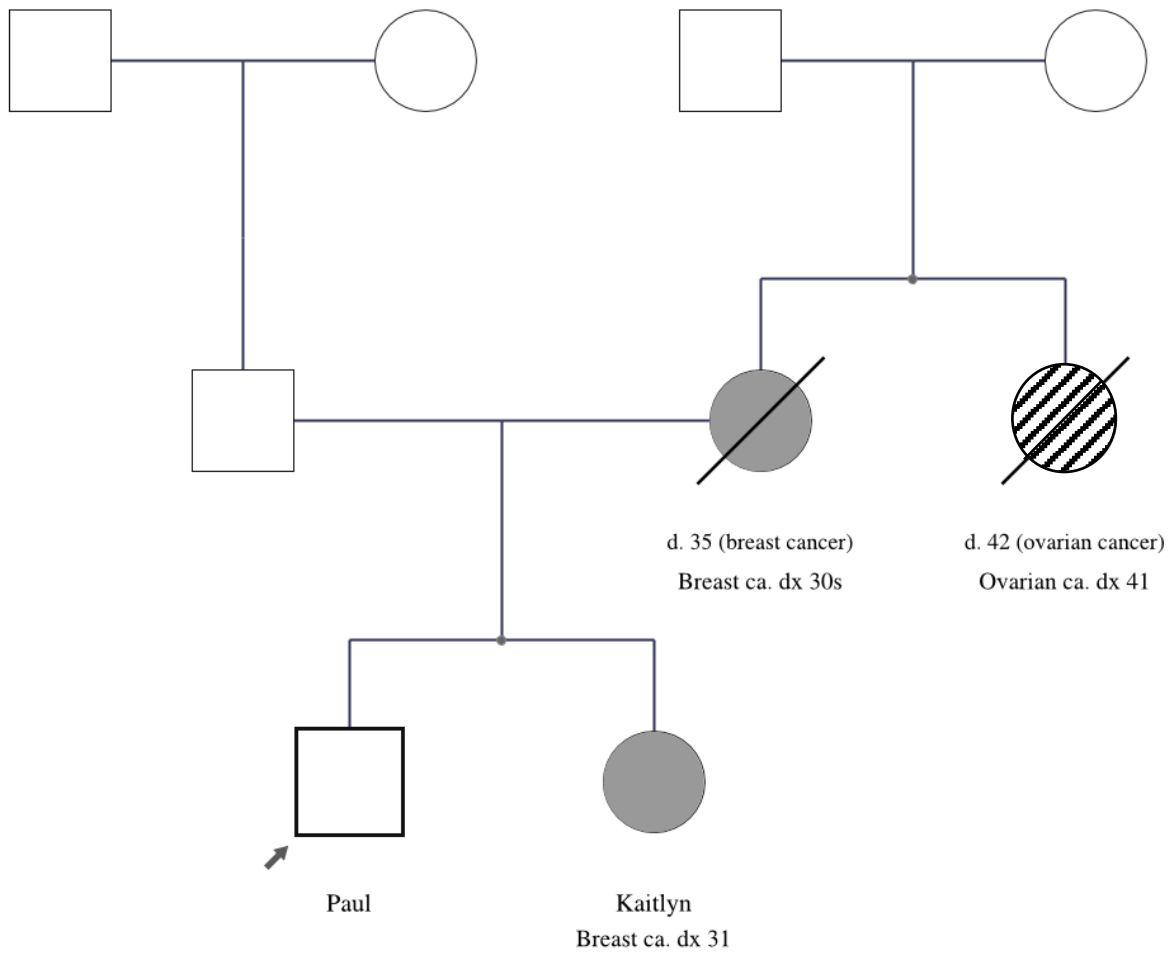


Table 2. Demographic characteristics of the participants.

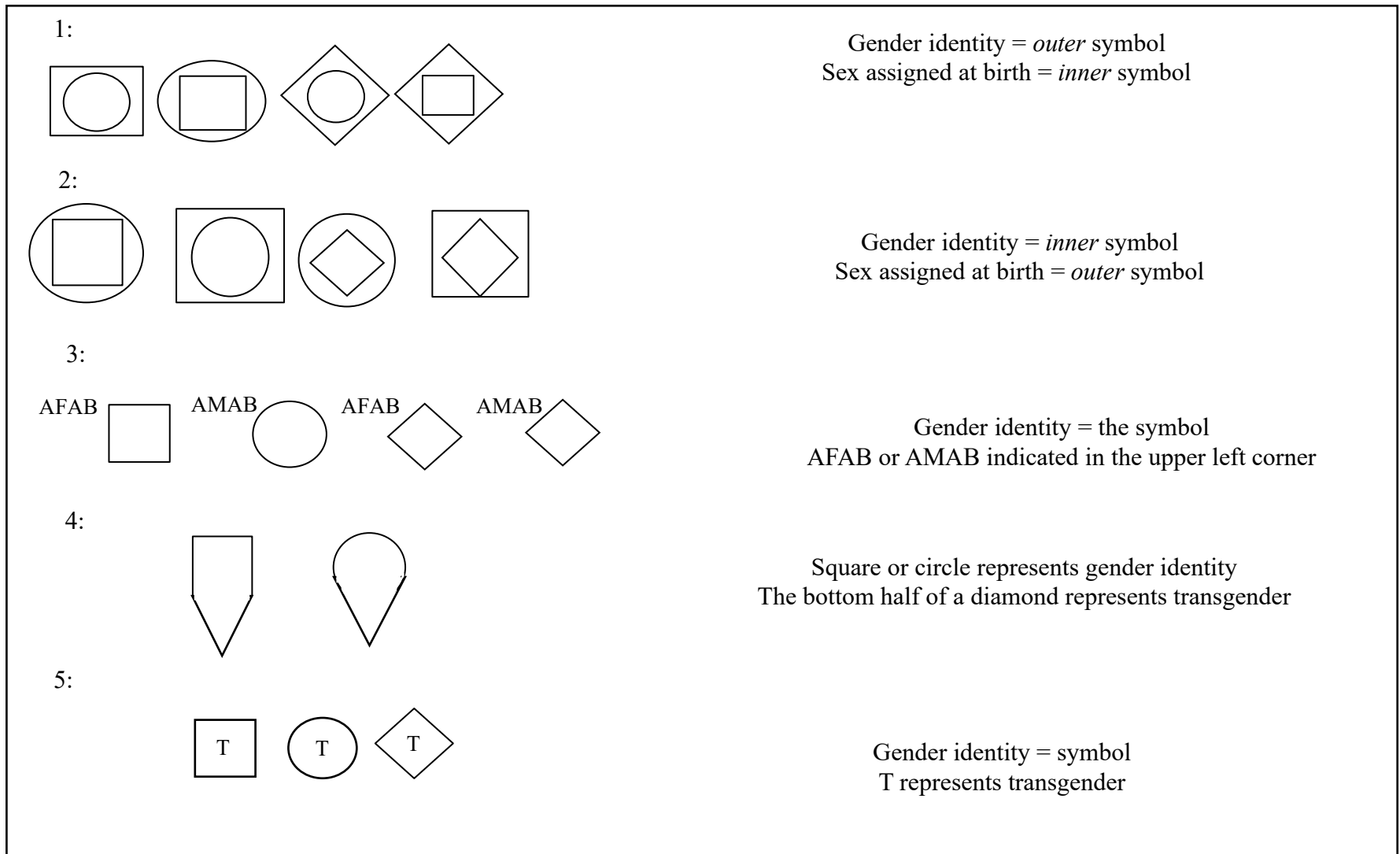
Participant Characteristic	N
Average age (range; median)	31.88 (22-62; 25)
Ethnicity	
White	8
Education level	
Some university	4
Bachelor's Degree	3
Master's Degree	1
Employed	8
Marital status	
Single	2
Common-law, in a relationship	4
Married	1
Separated	1
Median number of children (range)	0 (0-3)
Medical Interventions	
Hormone Treatment	6
Surgery	4

Table 3. Participants' gender identities.

Participant¹	Pronouns used at the time of study participation	Sex assigned at birth	Gender Identity	Gender Expression
Aaron	He, him, his	Female	Transmasculine	Male and gender non-conforming
Edward	He, him, his	Female	Transmasculine	Male
Mara	She, her, hers	Male	Transfeminine	Sometimes male, sometimes female
Nicholas	He, him, his OR They, them, theirs	Female	Transmasculine and non-binary	Male
Parker	They, them, theirs	Female	Transmasculine and non-binary	Neither male or female, and masculine
Ray	Ve, hir, hers	Female	Transmasculine	Gender non-conforming, female
Ruby	He, him, his	Male	Transfeminine	Male
Taylor	They, them, theirs	Female	Non-binary	Non-binary

¹Pseudonym provided by the participant or chosen by researchers.

Figure 3. Pedigree symbols suggested by participants. Where the diamond is used, it is used to represent non-binary or gender non-conforming individuals. Options 4 and 5 were excluded as possibilities by the researchers for practical reasons or due to ambiguity of no correlating representation for non-binary individuals.



AFAB – assigned female at birth

AMAB – assigned male at birth

Table 4. Recommendations for providing trans-inclusive genetic counseling.

- Forms and documents:
 - Provide clinic forms that allow for patients to indicate their preferred name, and select from or write in a variety of potential pronouns, gender identities and sex assigned at birth (see Bauer, Braimoh, Scheim, & Dharma, 2017 for suggestions for wording).
 - If sending out family history forms in advance of an appointment, include a question about whether they know of any family members whose birth sex does not align with their gender identity.
- Introductions:
 - Use your own pronouns in your greeting and introduction, i.e. “My pronouns are she/her. What are your pronouns?”
 - Ask each patient their preferred name.
 - Ask each patient about sex and gender, i.e. “What is your sex assigned at birth? What is your gender identity?”
- Pedigree Questions:
 - Preface the pedigree: “I will use shapes to represent you and your family members. It may be important for my evaluation to know both the gender identity and the sex assigned at birth of yourself and your family members so everyone is represented accurately on the pedigree. So, it would be really helpful if you could let me know as we go if there is anyone in the family whose birth sex and gender identity do not align.”
 - Check at the end of the pedigree: “Is there anyone in your family who identifies as transgender?” OR “Is there anyone in your family who has a different gender identity than the sex they were assigned at birth?”
- Throughout the session:
 - Using inclusive phrases e.g. “people who have a Y chromosome are at risk” or “individuals who are assigned male at birth are at risk” instead of “men are at risk”
 - If accidentally misgendering someone: apologize sincerely but briefly and continue with the error corrected.
 - Address privacy and confidentiality concerns: inform the patient about who may have access to the pedigree now or in the future. Some patients may not feel safe disclosing their own gender identity or that of a family member; to protect privacy, the genetic counselor could provide the option of symbolizing the individual differently on pedigrees provided to other healthcare providers or family members.

TRANS-INCLUSIVE GENETIC COUNSELLING SERVICES: RECOMMENDATIONS FROM THE TRANSGENDER COMMUNITY

Educational Document

SYMBOL USAGE IN GENETIC COUNSELLING

In genetic counselling, pedigrees are a visual representation of a family history and are used by genetic counsellors to provide a proper assessment of the inheritance pattern of disease in the family. Symbols are used in the pedigree to represent a patient and their family members. Creating pedigrees allows for a proper evaluation of the chance for the patient and their family members to develop a particular condition and for understanding family dynamics and/or social situation. The symbols currently used in genetic counselling are indicated below:



A square is used to represent **males**



A circle is used to represent **females**



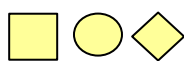
A diamond is used when gender is **not known** or not relevant



A diamond with a 'P' inside represents a **pregnancy**



A triangle is used to represent **miscarriages**



Coloured symbols are used to represent individuals **affected** with a particular condition.



Symbols with black circles inside them represent individuals who are **carriers** of the condition. They are typically unaffected by the condition.



Symbols with lines across them represent individuals who are **deceased**.

Transgender Symbols

One of the reasons we are doing this study is because there is no clear consensus on how best to represent transgender individuals on a pedigree. Current guidelines suggest that the diamond symbol can be used to represent **transgender** individuals. The guidelines, however, also suggest that a square or circle can be used, but it is not clear what this decision should be based on (e.g. sex assigned at birth or current gender identity).

It is important for genetic counsellors to know an individual's sex as assigned at birth as this also usually reflects their chromosomes (that is, if they have XX or XY chromosomes) to understand their chance for developing the condition or passing the condition on to future children. But it is also important to know about an individual's gender identity in order to appropriately address them.

Description: Trans-Inclusive Genetic Counselling

Version: May 25, 2018