

SPECIAL ISSUE

Beyond circles and squares: A commentary on updating pedigree nomenclature to better represent patient diversity

Allysa Tuite¹  | Michelle Dalla Piazza²  | Kristyn Brandi³  | Beth A. Pletcher¹ 

¹Division of Clinical Genetics, Department of Pediatrics, Rutgers New Jersey Medical School, Newark, NJ, USA

²Division of Infectious Disease, Department of Medicine, Rutgers New Jersey Medical School, Newark, NJ, USA

³Department of Obstetrics, Gynecology, and Women's Health, Rutgers New Jersey Medical School, Newark, NJ, USA

Correspondence

Allysa Tuite, Department of Pediatrics, Division of Clinical Genetics – Rutgers New Jersey Medical School, 90 Bergen Street, Suite 5400, Newark, NJ 07103 USA.
Email: allygo@njms.rutgers.edu

Abstract

With changes in our understanding of gender identity and disorders of sex differentiation (DSDs), as well as a need to promote medical care that appropriately reflects the intersectional personal identities of patients with respect to sex and gender, we explored possible modifications of pedigree nomenclature to better represent such patient diversity. There are currently no widely accepted standard symbols to simultaneously represent both gender identity and assigned sex at birth within a pedigree. Previous studies assessing perspectives from members of the transgender and gender non-binary (TGNB) community have highlighted the need for a unique symbol to represent non-binary individuals and better ways to represent core gender identities for gender minorities such as transgender individuals. In our experience we have encountered similar dilemmas with documentation for individuals with DSDs in terms of a lack of unequivocal symbolic representation within the pedigree. Here we propose three distinct symbols for gender identity combined with superscript symbols to represent sex assigned at birth, which we think may unequivocally represent TGNB individuals and patients with DSDs. It is clear that further research is needed to ensure that any proposed changes are acceptable by and respectful of all patients regardless of their gender identity and assigned sex at birth. We hope that further research will include focus groups and surveys to get broader input from gender minority stakeholders so that new standards can be developed and modified as we strive to meet the needs of our increasingly diverse patient population.

KEYWORDS

cultural competence, diversity, gender minorities, intersex, non-binary, pedigree nomenclature, transgender, underrepresented populations, attitudes, communication, community, cultural competence, disparities, education, family history, genetic counseling, genetics services, psychosocial, access

1 | INTRODUCTION

In a recent article published by Heather Barnes and colleagues (Barnes et. al., 2019), members of the transgender and gender non-binary (TGNB) community were invited to provide feedback about how best to promote trans-inclusive genetic services, using

easily understood pedigree symbols that accurately and respectfully reflect sex and gender. The survey participants specifically expressed dissatisfaction with the proposed diamond symbols for individuals who are TGNB. Participants reported that the diamond symbol lacked clarity, was not validating, and does not differentiate between trans-men, trans-women, and non-binary individuals.

The majority of respondents also felt that both assigned sex at birth (ASAB) and gender identity should be included within the pedigree. The majority of respondents given three possible scenarios preferred to have a single symbol representing their gender identity, with the option of an annotation of AFAB or AMAB to reflect sex assignment at birth (assigned female at birth and assigned male at birth, respectively).

It is evident that in order to provide culturally effective, patient-centered genetic care, we need a new paradigm to meet the needs of the TGNB community and must invite members of this community to partner with us to devise strategies to best represent gender identity and sex within a pedigree. Accurate representation will go far in helping all genetic providers to appropriately represent these attributes that are critical to optimal genetic counseling, risk assessment, and medical management.

As medical and genetic counseling educators, we too have grappled with possible ways to address this unmet need to best represent the diversity of our patients presenting for care with pedigree symbols. Over the past year, while awaiting updates to accepted nomenclature, we created a more inclusive pedigree modification for our internal use. We aimed to represent both gender identity and ASAB to depict more broadly and simply the histories of our patients in visual form. These symbols could potentially be used in standard pedigree-taking to easily assign each individual in a pedigree both a gender symbol and ASAB designation, with potential additional annotations if necessary to indicate laboratory findings. Patients who would most benefit from these modifications include those who identify as TGNB and/or individuals with disorders of sex differentiation (DSDs).

1.1 | A case for proposed pedigree nomenclature updates to promote sex and gender inclusion

1.1.1 | Limitations of the traditional approach

The traditional pedigree employs circles and squares to represent females and males, respectively. However, simple circles and squares cannot accurately reflect the intersectional personal identities of our patients with respect to sex and gender. In our view, given the health effects evident from both sex (ASAB—as determined at birth by visual inspection of the genitalia or by karyotype analysis, hormones, internal/external reproductive organs, and secondary sex characteristics) and gender (as determined by one's own self-knowledge, and which has significant social effects on health), accurate representation on a pedigree is critical for assessment of both genetic risks and the prediction of potential health outcomes based on social factors. Currently, there is no universally accepted manner by which to represent cisgender (gender identity and ASAB congruent), transgender (gender identity differs from ASAB), gender non-binary (gender identity on a spectrum or outside of the traditional gender binary) or intersex in the pedigree (GLAAD, 2011; UCSF Center of Excellence for Transgender Health, 2016).

As noted in previous studies regarding genetic counselors' experiences working with transgender populations, there has not been a consensus regarding optimal ways to identify transgender individuals within a pedigree (Barnes et al., 2019; Berro, Zayhowski, Field, Channaoui, & Sotelo, 2019; Zayhowski et al., 2019). The two most cited references regarding pedigree nomenclature include

	Identifies as girl/woman	Identifies as boy/man	Identifies as non-binary
Assigned female at birth	<p>Cis girl/woman</p>	<p>Trans boy/man</p>	<p>Non-binary</p>
Assigned male at birth	<p>Trans girl/woman</p>	<p>Cis boy/man</p>	<p>Non-binary, assigned male at birth</p>
Assigned intersex at birth	<p>Girl/woman, assigned intersex at birth</p>	<p>Boy/man, assigned intersex at birth</p>	<p>Non-binary, assigned intersex at birth</p>

FIGURE 1 Proposed updates for pedigree nomenclature

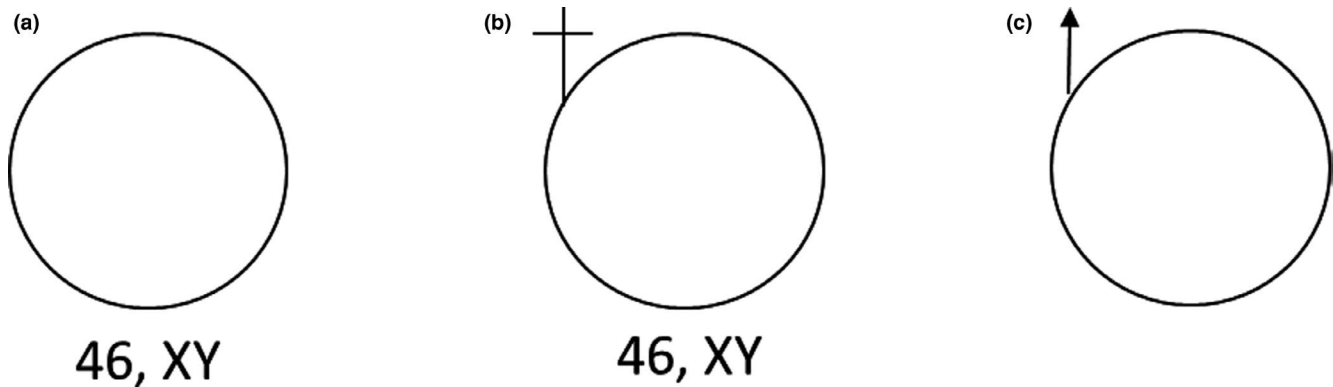


FIGURE 2 (a) Current nomenclature for woman with XY karyotype or transgender girl/woman; (b) proposed nomenclature for girl/woman, AFAB with known karyotype of XY due to XY sex reversal; (c) proposed nomenclature for transgender woman, AMAB

guidelines put forth by the National Society of Genetic Counseling Pedigree Task Force (Bennet, French, Resta, & Doyle, 2008), and most recently the National Comprehensive Cancer Center Network (NCCN)'s recommendations on how to represent transgender individuals in a pedigree (Provenzale et al., 2017).

The Pedigree Standardization Task Force of NSGC proposes documenting gender identity as shape, with the ASAB not specifically documented, and if known, the corresponding karyotype (e.g., 46, XY [ASAB male] or 46, XX [ASAB female]) noted below the symbol (Bennett et al., 2008). However, the use of the shape symbol for gender identity alone without additional information such as a karyotype or ASAB may be problematic or confusing. As an example, several DSDs may cause discrepancies between karyotype and ASAB. The NCCN proposes either a circle within a square representing a transgender man or a square within a circle representing a transgender woman. Attempts to represent a symbol within a symbol for transgender individuals are also confusing regarding the patient's current gender versus ASAB and may show insensitivity to a person's identity as a single core gender.

The current proposal by NSGC is for non-binary individuals to be denoted by a diamond. However, since the diamond can also represent gender not specified, such as with an ongoing pregnancy, it does not respectfully or accurately depict a non-binary individual. In a previous study, non-binary individuals expressed a need for a definitive symbol distinguishing between transgender men, transgender women, and non-binary individuals and that the use of the diamond 'did not feel validating' (Barnes et al., 2019). In such situations, our patients clearly state their gender is known, and that they identify as neither man nor woman. Equally problematic, we were unable to find a unique designation for assigned intersex at birth. Unfortunately, research regarding transgender, gender non-binary, and intersex patients' perspectives on symbolic representation within the pedigree has been limited (Barnes et al., 2019; Berro et al., 2019; Zayhowski et al., 2019). Barnes et al. (2019) propose to use a square/circle to represent gender identity with the annotation of 'AFAB' or 'AMAB' to indicate 'assigned female/male at birth'; however, they also proposed the continued use of a diamond to represent a non-binary individual with the

annotation AFAB/AFAB outside the symbol. Due to the multiple uses of a diamond in a pedigree, it would be imperative to create a unique symbol that would validate and depict non-binary gender identity. It benefits the community to provide respectful and accurate ways to represent these identifiers in the pedigree format.

1.1.2 | Our proposed changes

To address the issues outlined above, we have internally adopted the changes to the nomenclature as depicted in Figure 1. Similar to the NSGC guidelines, we use the circles and squares to designate gender identity, where a man is designated by a square, and a woman by a circle. We propose to use a new symbol, the inverted triangle, to signify a person who identifies as non-binary, questioning gender, or other genders. Our second proposal is to add small 'superscript symbols' in the upper left-hand corner of the gender identity symbol to represent the ASAB. Based on the easily identifiable symbols derived from astrologic signs for female and male, a cross represents AFAB, and an upward arrow represents AMAB. We are proposing the use of a lower case 'i' to represent assigned intersex at birth. In this way, the traditional shape primarily represents an individual's core gender identity; at the same time, the secondary ASAB symbol allows the provider to simultaneously account for a risk assessment based on both gender and ASAB. As with many cases of pedigree-taking, information such as karyotype is not easily inferred or distinguished.

This proposal addresses the pragmatic issues of other pedigree alternatives raised by Barnes et al. (2019) such as the difficulties to freehand a new symbol or alternate symbols that may involve shading or including information within the symbol. The inverted triangle is a unique symbol that is not currently being used in pedigree-taking. Our proposed changes also do not require any shading of symbols or additional writing or letters within or below the symbol. The addition of a superscript eliminates the need for 'AMAB' or 'AFAB' to be written below the pedigree symbol and would allow for more efficient and simple documentation. These proposed changes would allow for more organized pedigree-taking and streamline the pedigree-taking process by eliminating the need for extraneous or ambiguous symbols

or words below the symbol. If necessary or available, additional relevant medical information such as hormone status or secondary sex characteristics can be documented below the symbol as needed.

1.1.3 | Specific examples

In a medical genetic setting, these proposed changes in nomenclature would be helpful in distinguishing a transgender person from a person with a DSD. One example is a patient whose ASAB is female, identifies as girl/woman, and has a karyotype of 46, XY due to XY sex reversal. Given the current guidelines, the patient would be designated as seen in Figure 2a with a circle with '46, XY' written below. However, with the current guidelines, this could also represent a transgender girl/woman who has a known 46, XY karyotype. Our proposed update for this particular patient would be represented in Figure 2b with the cross signifying that the patient was AFAB, the circle signifying that the patient identifies as a girl/woman and '46, XY' which represents the patient's actual karyotype. Figure 2c would represent a transgender woman, with the arrow representing that this patient was AMAB, the circle representing that the patient is a woman, and the option to write '46, XY' below only if the patient's karyotype is known. Therefore, for the majority of transgender individuals, no karyotypic designation or additional nomenclature such as 'AFAB' or 'AMAB' would be added since the primary symbol and superscript symbol would suffice. This proposed update would allow for the subtle addition of a more accurate representation of individuals in pedigrees, covering the broad range of gender identities, ASAB, and, when necessary and available, cytogenetic results.

2 | CONCLUSION

The issue of gender and sex is important for understanding risks such as during cancer counseling and screening, reproductive risk assessment, and sex-based risk assessments for other medical concerns associated with genetic conditions. As healthcare providers, we recognize that many health risks are biological risks associated with one's ASAB. At the same time, understanding and accurately representing an individual's gender identity—whether cisgender, transgender, or an alternative gender—have important health implications from a biosocial perspective. Issues regarding sex and gender for gender minorities can be fraught with misunderstandings, and there is a need for all healthcare professionals to create a sense of belonging so that we can better meet the needs of our patients, not only with respect to their medical concerns, but also their psychosocial wellbeing. Being able to quickly and accurately represent individuals of all genders and sexes on a pedigree is one step closer to ensuring equitable treatment and advancing a truer understanding of each individual's concerns, health risks, and health needs.

We recognize the imperative for engaging with leaders in both the gender minority and intersex communities as well as the genetic counseling community to evaluate the appropriateness of these proposed symbols. For the transgender, non-binary, and intersex communities, it is critical to ensure that any proposed representations

are accurate, respectful, and comprehensive. For the genetic counseling community at large, we need to ensure that these proposed additions are feasible, accessible, and able to be readily incorporated into practice. Our next step is to develop focus groups and surveys for input from these stakeholders. We hope that our proposal will, at a minimum, invite more conversations about how best to represent the intersectional personal identities of our patients in a way that supports a patient-centered approach to care and genetic counseling, for our wonderfully diverse patients.

AUTHOR CONTRIBUTIONS

Mrs. Allysa Tuite made substantial contributions to the commentary conception, drafted and co-wrote the initial draft, revised the manuscript critically, and approved the final version. She agrees to be accountable for all aspects of the work, ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. Dr. Michelle Dalla Piazza conceptualized the initial pedigree designations in conjunction with Dr. Pletcher, reviewed and substantially revised the second draft of the commentary, and approved the final version. She agrees to be accountable for all aspects of the work, ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. Dr. Kristyn Brandi was integrally involved in the conceptualization of this commentary, reviewed and substantially revised the second draft of the commentary, and approved the final version. She agrees to be accountable for all aspects of the work, ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. Dr. Beth Pletcher conceptualized the initial pedigree designations in conjunction with Dr. Dalla Piazza, reviewed and substantially revised both drafts of the commentary, and approved the final version. She agrees to be accountable for all aspects of the work, ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

COMPLIANCE WITH ETHICAL STANDARDS

Conflict of interest

Mrs. Allysa Tuite, Dr. Michelle Dalla Piazza, Dr. Kristyn Brandi, and Dr. Beth A. Pletcher no conflicts of interest.

Human studies and informed consent

No human subjects data were collected in the development of this commentary manuscript.

Animal studies

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

ORCID

Allysa Tuite  <https://orcid.org/0000-0001-7703-3378>

Michelle Dalla Piazza  <https://orcid.org/0000-0003-3262-2368>

Kristyn Brandi  <https://orcid.org/0000-0002-5132-7308>

Beth A. Pletcher  <https://orcid.org/0000-0002-2839-0819>

REFERENCES

- Barnes, H., Morris, E., & Austin, J. (2019). Trans-inclusive genetic counseling services: Recommendations from members of the transgender and non-binary community. *Journal of Genetic Counseling, 00*, 1–12. <https://doi.org/10.1002/jgc4.1187>
- Bennett, R. L., French, K. S., Resta, R. G., & Doyle, D. L. (2008). Standardized human pedigree nomenclature: Update and assessment of the recommendations of the national society of genetic counselors. *Journal of Genetic Counseling, 17*(5), 424–433. <https://doi.org/10.1007/s10897-008-9169-9>
- Berro, T., Zayhowski, K., Field, T., Channaoui, N., & Sotelo, J. (2019). Genetic counselors' comfort and knowledge of cancer risk assessment for transgender patients. *Journal of Genetic Counseling, 00*, 1–10. <https://doi.org/10.1002/jgc4.1172>
- GLAAD (2011). GLAAD media reference guide-transgender. Retrieved from <https://www.glaad.org/reference/transgender>
- Provenzale, D., Gupta, S., Regenbogen, S. E., Hampel, H., Slavin, T. P., Hall, M. J. ... Ogba, N. (2017). National Comprehensive Cancer Network: Genetic/familial high-risk assessment: Colorectal, 3.2017.
- UCSF Center of Excellence for Transgender Health (2016). Terminology and definitions. Retrieved from <http://transhealth.ucsf.edu/trans?page=guidelines-terminology>
- Zayhowski, K., Park, J., Boehmer, U., Gabriel, C., Berro, T., & Campion, M. (2019). Cancer genetic counselors' experiences with transgender patients: A qualitative study. *Journal of Genetic Counseling, 28*(3), 641–653. <https://doi.org/10.1002/jgc4.1092>

How to cite this article: Tuite A, Dalla Piazza M, Brandi K, Pletcher BA. Beyond circles and squares: A commentary on updating pedigree nomenclature to better represent patient diversity. *J Genet Couns.* 2020;29:435–439. <https://doi.org/10.1002/jgc4.1234>