

# Establishing Gender-Affirming Genetic Counseling Services

Publication Date: Dec 6, 2023

## Collection Editor(s):

Lex N. Powers, MS, CGC

Genetic Counselor, Division of Craniofacial Medicine, Seattle Children's Hospital, Seattle, Washington, USA

Robin L. Bennett, MS, CGC, ScD Hon

Professor of Medicine, Division of Medical Genetics, Department of Medicine, University of Washington, Seattle, Washington, USA

## Introduction

Transgender (trans) describes an individual whose gender identity differs from the sex they were assigned at birth. In the U.S., approximately 2.6 million people (~1% of the population) identify as transgender and this number is predicted to increase with growing awareness and societal acceptance of human gender diversity. Individuals whose gender identity falls outside of the gender binary (i.e., not exclusively man or woman) may use various labels to describe their gender, including but not limited to: non-binary, gender non-conforming, and genderqueer. We use the umbrella term "gender-diverse" here to describe these identities. It is important to recognize that while many gender-diverse people identify as trans, others do not.

In contrast, intersex refers to a variety of conditions in which a person is born with sexual/reproductive anatomy that does not fit the binary sex categories of male and female. It is estimated that ~1.7% of the world population is born with an intersex trait. Today, Differences of Sex Development (DSD) is the accepted term used by the medical community to describe congenital conditions involving atypical development of chromosomal, gonadal, or anatomical sex; however, the intersex community has voiced objection to this term because it is pathologizing. While it is possible to be both trans and intersex, they are not the same. These communities may confront similar (and often intertwined) challenges, as evidenced in this collection, but each have their own unique experiences of oppression.

Transgender, gender-diverse, and intersex (TGDI) individuals face higher rates of adverse health outcomes and poorer access to care compared to the general population. One significant barrier to care for these communities is interpersonal and systemic discrimination by providers and medical institutions, resulting in medical distrust and avoidance of healthcare services. For example, in a [study](#) conducted by the Center for American Progress (CAP) in 2020, nearly half of trans and gender-diverse respondents reported having experienced some form of mistreatment by a healthcare provider (including care refusal, misgendering, and verbal/physical abuse) and ~40% reported postponing or avoiding getting preventative cancer screenings due to prior discrimination. A [systematic review](#) published the same year demonstrated that intersex people experience a higher incidence of anxiety, depression and psychological distress due to stigmatization and discrimination by healthcare providers. These health inequities have prompted the genetic counseling field to broaden research and dialogue focused on determining and implementing best practices in caring for TGDI individuals.

As healthcare providers with expertise in the complexity of sex determination, genetic counselors have a professional responsibility to address the historical mistreatment of TGD patients and to correct the misconception that sex and gender are strictly binary. This collection serves as a starting point for understanding the TGD patient experience, existing barriers to providing TGD-inclusive care, and current gender-affirming genetic counseling practices. This is not an exhaustive list and literature on this subject continues to be published. Preferred language around sex, gender, and sexuality is also constantly evolving. To best serve our TGD patients, readers are encouraged to prioritize self-education and commit to life-long learning on this topic.

## Commentaries on TGD-Inclusive Care

- Bardach, P. (2023, June 14). [An urgent call for genetics organizations to support transgender and intersex communities](#). NSGC-Perspectives.
- Berro, T., & Zayhowski, K. (2023). [Toward depathologizing queerness: An analysis of queer oppression in clinical genetics](#). *Journal of Genetic Counseling*. Advance online publication.
- Di Gioacchino, V., Essendrup, A., Galasinski, S., Gilats, M., & Neufeld-Kaiser, W. (2022, April 7). [Gender inclusivity in the genetics lab](#). NSGC-Perspectives.
- Llorin, H., & Zayhowski, K. (2023). [The erasure of transgender and intersex identities through fetal sex prediction and genetic essentialism](#). *Journal of Genetic Counseling*, 32(5), 942–944.
- Bedrick, B. S., Fruhauf, T. F., Martin, S. J., & Ferriss, J. S. (2021). [Creating breast and gynecologic cancer guidelines for transgender patients with BRCA mutations](#). *Obstetrics and Gynecology*, 138(6), 911–917.

## TGD Patient Experiences in Genetic Counseling

- Barnes, H., Morris, E., & Austin, J. (2020). [Trans-inclusive genetic counseling services: Recommendations from members of the transgender and non-binary community](#). *Journal of Genetic Counseling*, 29(3), 423–434.
- Rolle, L., Zayhowski, K., Koeller, D., Chiluiza, D., & Carmichael, N. (2021). [Transgender patients' perspectives on their cancer genetic counseling experiences](#). *Journal of Genetic Counseling*, 31(3), 781–791.
- Valentine, R., Mills, R., Nichols, T., & Doyle, L. (2023). [Disclosure and comfort during genetic counseling sessions with LGBTQ+ patients: An updated assessment](#). *Journal of Genetic Counseling*, 32(4), 833–845.

## Intersex Patient Perspectives

- Haghighat, D., Berro, T., Torrey Sosa, L., Horowitz, K., Brown-King, B., & Zayhowski, K. (2023). [Intersex people's perspectives on affirming healthcare practices: A qualitative study](#). *Social Science & Medicine*, 329, Article 116047.
- Crocetti, D., Monro, S., Vecchietti, V., & Yeadon-Lee, T. (2021). [Towards an agency-based model of intersex, variations of sex characteristics \(VSC\) and DSD/DSD health](#). *Culture, Health & Sexuality*, 23(4), 500–515.
- Richardson, J. P., Ahlawat, N., Riggan, K. A., Close, S., & Allyse, M. A. (2022). [Experiences of individuals receiving a sex chromosome multisomy diagnosis](#). *Journal of Community Genetics*, 13(6), 619–628.

## Genetic Counselor Experiences in Caring for TGD Patients

- Berro, T., Zayhowski, K., Field, T., Channaoui, N., & Sotelo, J. (2020). [Genetic counselors' comfort and knowledge of cancer risk assessment for transgender patients](#). *Journal of Genetic Counseling*, 29(3), 342–351.
- Ruderman, M., Berro, T., Torrey Sosa, L., & Zayhowski, K. (2021). [Genetic counselors' experiences with transgender individuals in prenatal and preconception settings](#). *Journal of Genetic Counseling*, 30(4), 1105–1118.
- 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.

## Practice Resources, Recommendations, and Tools

- Bennett, R. L., French, K. S., Resta, R. G., & Austin, J. (2022). [Practice resource-focused revision: Standardized pedigree nomenclature update centered on sex and gender inclusivity: A practice resource of the National Society of Genetic Counselors](#). *Journal of Genetic Counseling*, 31(6), 1238–1248.
- Huser, N., Hulswit, B. B., Koeller, D. R., & Yashar, B. M. (2022). [Improving gender-affirming care in genetic counseling: Using educational tools that amplify transgender and/or gender non-binary community voices](#). *Journal of Genetic Counseling*, 31(5), 1102–1112.
  - To access the most up-to-date modules discussed in this study (Amplify/Sprouted: Evolved) enter the password Amplify2023 [here](#).
- Powers, L. N. (2023, February 22). [Gender-inclusive language guide for genetic counselors, providers, and instructors](#) [Document]. Microsoft OneDrive Live.
- Reckhow, J., Kula, H., & Babayev, S. (2023). [Fertility preservation options for transgender and nonbinary individuals](#). *Therapeutic Advances in Endocrinology and Metabolism*, 14.
- Hodan, R., Rodgers-Fouche, L., Chittenden, A., Dominguez-Valentin, M., Ferriss, J., Gima, L., Hamnvik, O. R., Idos, G. E., Kline, K., Koeller, D. R., Long, J. M., McKenna, D., Muller, C., Thoman, M., Wintner, A., Bedrick, B. S., & Collaborative Group of the Americas on Inherited Gastrointestinal Cancer. (2023). [Cancer surveillance for transgender and gender diverse patients with Lynch syndrome: A practice resource of the Collaborative Group of the Americas on Inherited Gastrointestinal Cancer](#). *Familial Cancer*, 22(4), 437–448.
- von Vaupel-Klein, A. M., & Walsh, R. J. (2021). [Considerations in genetic counseling of transgender patients: Cultural competencies and altered disease risk profiles](#). *Journal of Genetic Counseling*, 30(1), 98–109.

## Suggested Citation

Powers, L. N., & Bennett, R. L. (2023). Establishing gender-affirming genetic counseling services. In *ELSIhub Collections*. Center for ELSI Resources and Analysis (CERA).