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REVIEW ARTICLE

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Patient care practices for LGBTQ+ individuals in clinical genetics: A scoping review

Kimberly Zayhowski[1,2] | Kayla Horowitz[3] | Molly Bostrom[4] | Kathleen F. Mittendorf[5] | Megan Kocher[4] | Jehannine (J9) Austin[6,7] | Ian M. MacFarlane[2]

[1]Department of Obstetrics and Gynecology, Boston University Chobanian and Avedisian School of Medicine, Boston, Massachusetts, USA

[2]Department of Genetics, Cell Biology, and Development, University of Minnesota, Minneapolis, Minnesota, USA

[3]Service de Génétique Humaine, Cliniques Universitaires Saint-Luc, Brussels, Belgium

[4]Libraries, University of Minnesota, Minneapolis, Minnesota, USA

[5]Lavender Spectrum Health, Vancouver, WA, USA

[6]Department of Psychiatry, Faculty of Medicine, University of British Columbia, Vancouver, British Columbia, Canada

[7]Department of Medical Genetics, Faculty of Medicine, University of British Columbia, Vancouver, British Columbia, Canada

Correspondence
Kimberly Zayhowski, Department of Obstetrics and Gynecology, Boston University Chobanian and Avedisian School of Medicine, Boston, MA, USA.
Email: kzayhows@bu.edu

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Abstract

Individuals who are LGBTQ+ (Lesbian, Gay, Bisexual, Transgender, Queer/Questioning, and/or have a sexual orientations and/or gender identity beyond cisheteronormative conceptions) face systemic barriers to healthcare, leading to significant health inequities. To address these challenges, genetic providers must better understand and inclusively address LGBTQ+ patient needs. This scoping review aims to map the current landscape of genetic care practices and their inclusivity toward LGBTQ+ individuals. We conducted a systematic search of databases, including Ovid MEDLINE, PsycINFO, and Web of Science, identifying 65 relevant articles focused on LGBTQ+ patient experiences and care practices within genetic healthcare services. Our thematic analysis of the articles highlights three major themes: exclusionary clinical environments and tools, provider biases and educational needs, and patient-reported barriers in accessing genetic services. Many articles underscored the importance of inclusive language and criticized the conflation of sex, sex chromosomes, and gender. A significant focus was on cancer care for transgender and gender-diverse individuals, revealing a need for more data on the effects of gender-affirming care on cancer risk assessment. Moreover, genetic counselors often report insufficient training in LGBTQ+ health needs, contributing to biases and knowledge gaps. Despite increased awareness among providers of the need for inclusive care, LGBTQ+ patients encounter substantial barriers, including medical distrust and limited family health history, which may deter them from disclosing their identities due to the risk of discrimination. This review calls for standardized data collection practices regarding sex-related variables, gender modality, and sexual orientation, alongside specialized training programs for providers. By emphasizing critical areas for research, policy changes, and education, we aim to promote equitable, patient-centered genetic services for LGBTQ+ communities.

KEYWORDS
genetic counseling, genetics services, health inequity, LGBTQ+, scoping review, sexual and gender minority, transgender

1 | INTRODUCTION

Approximately 7.1% of adults in the United States openly identify as part of the LGBTQ+ (Lesbian, Gay, Bisexual, Transgender, Queer/Questioning, and individuals whose sexual orientation and/or gender modality/identity fall outside of cisheteronormative conceptions) community (Gallup, 2022). Table 1 describes the rationale we used surrounding language usage in this article. Representation of LGBTQ+ identities is higher among younger generations, with about one in five adults in Generation Z (defined as being born between 1997 and 2003) identifying as LGBTQ+, signaling a societal shift toward increased acceptance and visibility (Gallup, 2022; Herman et al., 2021). Despite recent progress, LGBTQ+ individuals continue to endure marginalization and medical pathologization. Inequities arising from social and structural barriers significantly exacerbate adverse health outcomes among LGBTQ+ individuals, such as those documented in adolescent medicine, sexual and reproductive health, mental well-being, and cardiovascular- and cancer-related conditions (Meyer, 2003; National Academies of Sciences, Engineering, and Medicine (NASEM), 2020; National Center for Chronic Disease Prevention and Health Promotion, 2019; National Institutes of Health (NIH), 2023; Scheim et al., 2022, 2024).

Systemic barriers to quality healthcare access for LGBTQ+ patients and persistent health inequities in this population underscore the critical need for the development and implementation of culturally responsive care practices (NASEM, 2020; National Institutes of Health, Sexual & Gender Minority Research Working Group of the Council of Councils, 2023; NIH, 2023; Scheim et al., 2022, 2024; Venetis et al., 2016). Healthcare providers and care delivery frameworks frequently assume cisgender and heterosexual status, exacerbating barriers to care by affecting the apparent safety of LGBTQ+ identity disclosure (Bergeron & Senn, 2003; Sequeira et al., 2020; Valentine et al., 2023). Binary frameworks of sex and gender in healthcare, reflected in language, documentation, and standardized practices, further marginalize LGBTQ+ identities and hinder effective communication, leading to inadequate care (Jamal et al., 2024). Healthcare providers often lack the training or resources to meaningfully engage LGBTQ+ patients, resulting in miscommunication and missed opportunities for tailored support (Yu et al., 2023). LGBTQ+ individuals experience sexual and gender modality-related healthcare needs (Ashley et al., 2024), yet these factors are frequently under-researched and under-addressed in genetic healthcare (Berro & Zayhowski, 2023; von Vaupel-Klein & Walsh, 2021).

While existing literature and reviews have delved into primary care, mental health, sexual health, and provider competency training (Charest et al., 2016; McNair & Hegarty, 2010; Moagi et al., 2021; Yu et al., 2023), genetics-specific research has been overlooked (Berro & Zayhowski, 2023; Jamal et al., 2024). The consistent exclusion of LGBTQ+ populations and considerations from genetic research and care delivery resources suggests a key area for future research and care delivery focus. This scoping review maps the current LGBTQ+ genetic services literature landscape to guide improved patient care practices and guidelines and to inform future LGBTQ+ inclusive research.

What is known about this topic

Existing literature indicates inequities in access to and quality of healthcare services for LGBTQ+ populations, emphasizing the importance of inclusive practices and further investigation into ways to remove systemic barriers to care. Emerging research on patient care practices for LGBTQ+ individuals in genetic counseling and genetic patient care highlights the need for tailored approaches to address the healthcare needs of this population.

What this paper adds to the topic

This scoping review provides a comprehensive assessment of the current knowledge landscape of genetic counseling and genetic patient care for LGBTQ+ individuals. Our knowledge synthesis identifies gaps, areas for improvement, and opportunities for future research related to inclusive and culturally responsive healthcare practices for LGBTQ+ populations.

2 | METHODS

We used the scoping review framework outlined in the Joanna Briggs Institute (JBI) Manual for Evidence Synthesis (Peters et al., 2022) to develop a review protocol. We adhered to the Preferred Reporting Items for Systematic Reviews and Meta-Analyses extension for Scoping Reviews (PRISMA-ScR) guidelines (Tricco et al., 2018) for results reporting. The protocol was preregistered on the Open Science Framework before search initiation (https://osf.io/vdq8e).

2.1 | Eligibility criteria

Peer-reviewed articles were included if they discussed LGBTQ+ patients or specific LGBTQ+ subgroups (e.g., gay individuals, transgender individuals, etc.) in genetic services. Inclusion criteria encompassed articles examining patient care practices, protocols, or interventions tailored to LGBTQ+ individuals, as well as publications discussing LGBTQ+ patient experiences and outcomes in the genetic healthcare services context. Articles were excluded if they were unrelated to LGBTQ+ patients or clinical genetics, exclusively focused on heterosexual or cisgender populations, or were not available in English. Articles solely related to intersex variations/variations of sex characteristics (VSC) were excluded, as the authors deemed this topic worthy of its own review due to the genetic nature of many VSC. Similarly, articles investigating genetic associations with LGBTQ+ identities were excluded, as these have extensive ethical implications that warrant a separate review.

TABLE 1 Usage and rationale for language choices in this review.

Term	Used to…	Rationale
LGBTQ+ (Lesbian, Gay, Bisexual, Transgender, Queer/Questioning, and/or have a sexual orientations and/or gender identity beyond cisheteronormative conceptions)	Encompass the entire spectrum sexual orientations and gender identities that fall under the scope of this article	Where possible, we have used identity-centric language to avoid further minoritizing individuals through language choices like "sexual and gender minority," which define them solely in relation to cisgender and/or heterosexual individuals. The National Institutes of Health (NIH) advocates for the use of sexual and gender minority in the context of health research and LGBTQI+ in the context of identity-focused applications like patient-facing materials (NIH, 2023). However, the experience of at least one author in various community focus groups in other research applications indicates that this language is seen as acting to other, medicalize and/or even pathologize sexual orientations and gender modalities that fall outside the bounds of cisheternormative ideals (Ashley et al., 2024). Because the authors believe that the scientific community should not be positioned as an objective outsider situated within cisheterosexual norms and that *all* scientific communication, including research articles, should have a community audience, we have used these identity-centric terms. As reviewed in the methods, we have not included intersex individuals and individuals with variations of sex characteristics in the scope of this review, as the literature base for this population is extensive and warrants a separate review
LGBQ+ (Lesbian, Gay, Bisexual, Queer/Questioning, and/or have a sexual orientations and/or gender identity beyond cisheteronormative conceptions)	Encompass sexual orientations that fall outside of societal heteronormative ideals	
TGD (transgender and gender-diverse)	Encompass gender modalities that fall outside of societal cisnormative ideals	
Gender identity	Refer to how a given citation termed a person's internal sense of self in relation to the social construct of gender and when gender modality could not be accurately applied to the cited article content (see below)	We have chosen to use the phrase gender identity as it was used in specific literature to conceptualize and measure a person's sense of gender at a given time, and we could not make an accurate statement utilizing the updated terminology of *gender modality* (see below)
Gender modality	Refer to the relationship between a person's gender identity and the gender they were assigned at birth	Where relevant in cited literature, our introduction, and our discussion, we have used the updated language of "gender modality" (Ashley et al., 2024), which refers to the relationship between a person's gender identity and the gender they were assigned at birth. Gender modality allows for classifications of cisgender and transgender, but also offers expansion beyond a "cis/trans" binary; the goal of this language change is greater inclusion of nonbinary people (especially those who are agender), some intersex people, and some gender creative people—as well as greater inclusion of gender experiences beyond those culturally situated within Eurocentric societies

2.2 | Information sources and search

Search strategies were developed in collaboration with the team's subject experts (KZ, KH, KM, JA, and IM) and librarians (MB and MK) using the databases Medline via Ovid, APA PsycInfo via Ovid, CINAHL Ultimate via EBSCOhost, Embase Classic via Ovid, and Scopus. Complete search strategies for each database are available in Appendix S1. The searches were conducted with no restrictions on publication date to ensure a comprehensive retrieval of relevant literature. They were originally run on November 30, 2023, with updates run on June 26, 2024, and October 17, 2024, to ensure results were as current as possible. On October 23, 2024, we added the additional terms "genomic" and "clinical genetics" which were missing from the original search terms but surfaced during the screening process, and newly retrieved articles were reviewed following the procedures above. The authors enhanced the search by conducting supplemental searches, such as manually screening references from included articles and contacting experts.

2.3 | Article selection

The Covidence web software facilitated article selection. After removing duplicates, titles and abstracts were independently screened by two reviewers (KZ and KH), with conflicts resolved through discussion with a third party (IM). Articles authored by team members underwent review by individuals within the study team who were not co-authors of those articles. Full-text screening of eligible titles

and abstracts was conducted independently (KZ and KH), with conflicts resolved via consensus.

2.4 | Data extraction

A data extraction form was developed based on study aims and piloted for reliability, with iterative refinement to ensure all pertinent data were captured. Two reviewers (KZ and KH) performed data extraction with cross-verification for accuracy. Extracted data were entered into Covidence for further analysis.

2.5 | Risk of bias assessment

Given the scoping nature of the review, no formal risk of bias assessment was conducted – consistent with JBI methods and PRISMA-ScR recommendations.

2.6 | Synthesis of results

Data from Covidence were exported into Excel for further analysis, data categorization, and organization. Extracted data were synthesized using descriptive analysis, including frequencies, to elucidate key findings. KZ and KH employed reflexive thematic analysis to conceptualize themes, with regular input from other team members to refine and validate results. Reflexive thematic analysis practices result in an inherently subjective knowledge generation process; we recognize that author positionalities influenced data interpretation. The author team comprises individuals with expertise in genetic counseling, research methodologies, and LGBTQ+ healthcare. Most authors are LGBTQ+ community members.

Our analysis followed the framework outlined by Braun and Clarke (2021), involving several key phases to ensure a systematic approach to data synthesis. Initially, we familiarized ourselves with the dataset through multiple readings, noting preliminary insights regarding LGBTQ+ experiences in genetic services, and conducting data extraction. During the coding phase, we employed inductive methods, developing initial codes based on recurring themes. We collaboratively reviewed and refined these themes with the study team, ensuring accuracy and relevancy. Each theme was further defined and named for clarity and specificity. Throughout writing, we iteratively refined the themes and codes to integrate new insights.

3 | RESULTS

As outlined in the PRISMA diagram (Figure 1), the screening process resulted in the evaluation of 1253 references, with 230 duplicates flagged by the Covidence tool and an additional four duplicates identified manually. Comprehensive title and abstract review of the remaining 1019 articles led to the exclusion of 923 articles that did not meet inclusion criteria. Subsequently, full-text review of 95 articles resulted in the exclusion of 30 articles; 8 articles were not oriented toward LGBTQ+ health-related needs, 6 were not focused on clinical genetics, and 4 dealt with fertility without substantial genetic relevance. Additionally, 5 articles did not address both clinical genetics and LGBTQ+ themes, 4 were not in English, 2 were centered on intersex topics, and 1 focused on research to find LGBTQ+ identity-associated genes. This process led to the inclusion of 65 articles for data extraction and synthesis.

3.1 | Article characteristics

Table 2 depicts the characteristics of each included article, along with extracted themes. Included articles exhibited a broad range of types and methodologies, including 14 commentaries, 13 quantitative analyses, nine qualitative analyses, nine case reports and case series, nine review articles, seven mixed-methods analyses, and three professional guidelines. For the purpose of this scoping review, manuscripts were labeled as commentaries if they were opinion pieces discussing issues or viewpoints without systematic analysis or comprehensive review of the literature. In contrast, we labeled articles as reviews if they provided structured summaries of existing research, aiming to synthesize findings and identify gaps. One article could not be labeled as one of these article types; Ernst et al. (2023) described the development of community-based educational modules. Thirty articles centered on cancer genetics, 14 on reproductive genetics, four on research methodologies, and two on laboratory practices, while 18 explored broader general genetic practices topics. Notably, four articles had a specialty focus: neurology (Bayram et al., 2024), cardiology (von Vaupel-Klein & Walsh, 2021), pediatric ophthalmology (Karmouta et al., 2023), and pharmacogenomics (Sehgal, 2023). The sum of these numbers exceeds 65, as six articles were multiply categorized. In terms of the predominant LGBTQ+ subgroup addressed in the articles, 44 focused on the healthcare needs of transgender and gender-diverse individuals (those whose gender identity does not align with cisnormative conceptions; TGD), 11 examined how sexual orientation impacts care, and 10 covered both sexual orientation and gender identity. Although articles solely focused on intersex individuals were excluded, several included articles explicitly acknowledged considerations for individuals with intersex variations/VSC or included participants who identify as intersex (Baecher-Lind et al., 2023; Bennett et al., 2022; Berro & Zayhowski, 2023; Bland et al., 2024; Burzynski et al., 2024; Cortina et al., 2024; Dusic et al., 2024; Jamal et al., 2024; Kamoun et al., 2023; Llorin et al., 2024; Llorin & Zayhowski, 2023; Saunders et al., 2024; Stevens et al., 2023; Tuite et al., 2020; Tyrie et al., 2024; Valentine et al., 2023; von Vaupel-Klein & Walsh, 2021). Not all articles were exclusively focused on LGBTQ+ issues, but these classifications reflect the articles' primary LGBTQ+-related content. Most included articles were published recently, with 48 out of 65 appearing between 2020 and 2024.

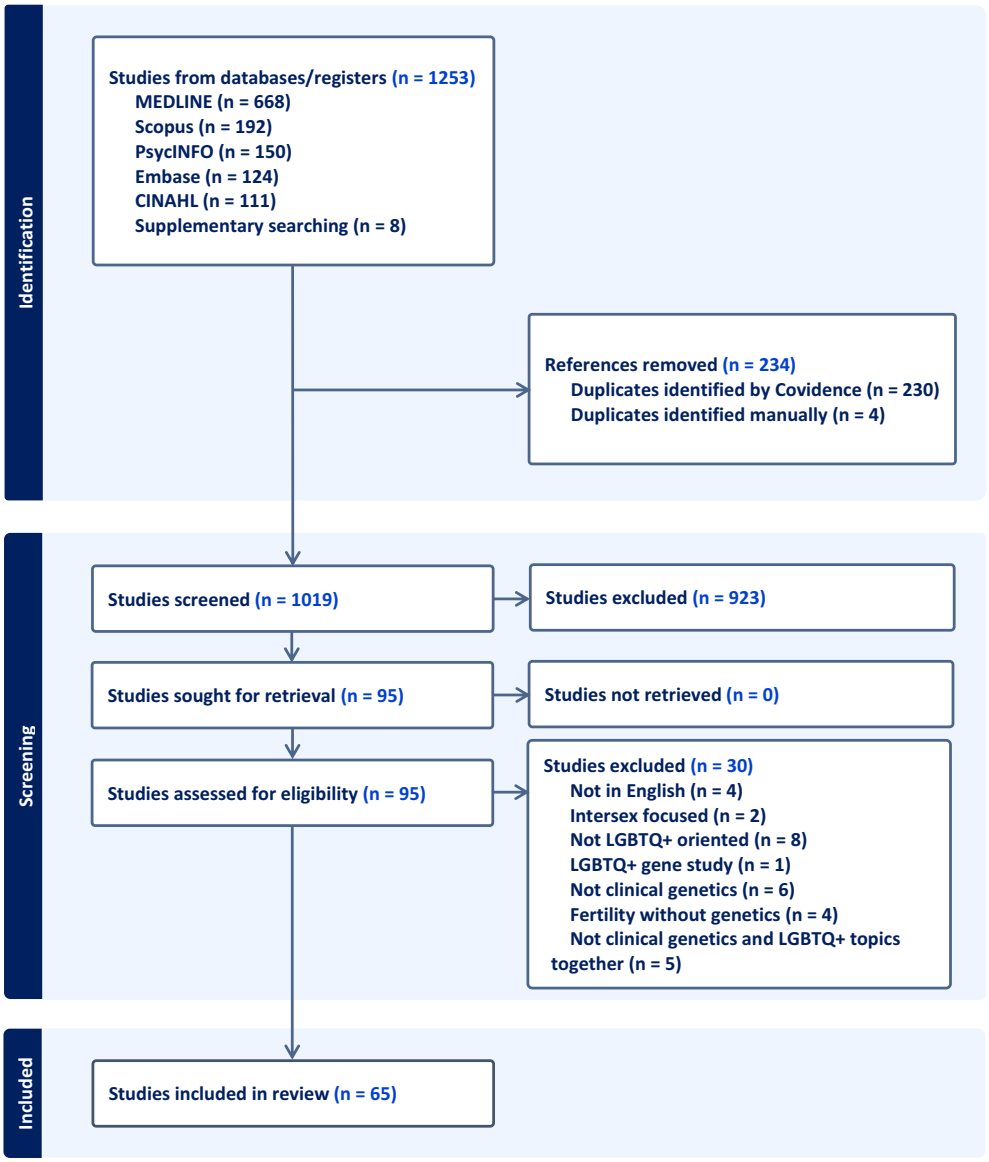

FIGURE 1 PRISMA diagram.

3.2 | Thematic analysis

Our thematic analysis synthesized findings from the included articles, emphasizing key areas for improvement in genetic services delivery for LGBTQ+ individuals. The thematic analysis is arranged into three overarching themes: (1) exclusionary clinical environments and tools, (2) genetic providers biases and educational needs, and (3) patient-reported barriers to genetic services.

3.2.1 | Exclusionary clinical environments and tools

We further delineated the overarching theme of exclusionary clinical environments and tools into specific clinical and content areas, including: (1) clinical tools, (2) reproductive environments, (3) conflation of sex, gender, and sex chromosomes, (4) cancer care for LGBQ+ individuals, (5) cancer genetic services for TGD patients, and (6) emerging areas of interest.

3.2.1.1 | *Clinical tools*

Identified articles criticized existing clinical tools in genetic counseling for lack of LGBTQ+ inclusivity, particularly in representing diverse gender identities and familial structures. Many articles highlighted the lack of standardized pedigree symbols and nomenclature to accurately represent gender diversity, either as a primary focus (Barnes et al., 2020; Bennett et al., 2022; Resta et al., 2023; Sheehan et al., 2020; Tuite et al., 2020) or as part of broader discussions (Baecher-Lind et al., 2023; Berro et al., 2020; Berro & Zayhowski, 2023; Coad et al., 2021; Giblin et al., 2023; Mahon, 2023; Ruderman et al., 2021; von Vaupel-Klein & Walsh, 2021; Zayhowski et al., 2019). Several articles emphasized that using gender-inclusive language and adopting flexible pedigree nomenclature more

TABLE 2 Article characteristics.

Article	Article type	Specialty focus[a]	Focus: LGBTQ+, LGBQ+, or TGD[b]	Themes extracted
Angelo et al. (2022)	Quantitative analyses	General practice	TGD	Patient reported barriers to genetic services
Antony et al. (2024)	Review	Cancer	TGD	Cancer genetic services for TGD patients
Atkins (2008)	Qualitative analyses	Reproductive	LGBQ+	Reproductive environments; Patient reported barriers to genetic services
Baecher-Lind et al. (2023)	Commentary	Reproductive	TGD	Clinical tools; Reproductive environments
Barnes et al. (2020)	Qualitative analyses	General practice	TGD	Clinical tools; Patient reported barriers to genetic services
Baylis (2018)	Commentary	Reproductive	LGBQ+	Reproductive environments
Bayram et al. (2024)	Quantitative analyses	Research; Other	LGBTQ+	Emerging areas of interest; Patient reported barriers to genetic services
Bennett et al. (2022)	Professional guideline	General practice	LGBTQ+	Clinical tools
Berro et al. (2020)	Quantitative analyses	Cancer	TGD	Clinical tools; Cancer genetic services for TGD patients; Genetic providers' biases and educational needs
Berro and Zayhowski (2023)	Review	General practice	LGBTQ+	Clinical tools; Reproductive environments; Conflation of sex, gender, and sex chromosomes; Cancer genetic services for TGD patients
Bland et al. (2024)	Case report	Cancer; Research	TGD	Clinical tools; Cancer genetic services for TGD patients
Burnett et al. (1999)	Quantitative analyses	Cancer	LGBQ+	Cancer care for LGBQ+ individuals; Patient reported barriers to genetic services
Burzynski et al. (2024)	Mixed analyses	Reproductive	TGD	Reproductive environments; Conflation of sex, gender, and sex chromosomes; Genetic providers' biases and educational needs
Cavaliere and Palacios-González (2018)	Commentary	Reproductive	LGBQ+	Reproductive environments
Chen et al. (2017)	Mixed analyses	General practice	LGBQ+	Patient reported barriers to genetic services
Coad et al. (2021)	Review	Cancer	TGD	Clinical tools; Cancer genetic services for TGD patients
Colebunders et al. (2014)	Case report	Cancer	TGD	Cancer genetic services for TGD patients
Corman et al. (2016)	Case report	Cancer	TGD	Cancer genetic services for TGD patients
Cortina (2023)	Commentary	Cancer	TGD	Cancer genetic services for TGD patients

TABLE 2 (Continued)

Article	Article type	Specialty focus[a]	Focus: LGBTQ+, LGBQ+, or TGD[b]	Themes extracted
Cortina et al. (2024)	Quantitative analyses	Cancer	TGD	Cancer genetic services for TGD patients
Daly et al. (2023)	Professional guideline	Cancer	TGD	Clinical tools; Cancer genetic services for TGD patients
Durfy et al. (1999)	Quantitative analyses	Cancer	LGBQ+	Cancer care for LGBQ+ individuals; Patient reported barriers to genetic services
Dusic et al. (2024)	Commentary	Laboratory	TGD	Clinical tools; Conflation of sex, gender, and sex chromosomes
Eckhert et al. (2023)	Quantitative analyses	Cancer	LGBTQ+	Cancer care for LGBQ+ individuals; Cancer genetic services for TGD patients
Ernst et al. (2023)	Other	General practice	TGD	Genetic providers' biases and educational needs
Friedman and Papagiannis (2024)	Case report	Cancer	TGD	Cancer genetic services for TGD patients
Giblin et al. (2023)	Review	Cancer	TGD	Clinical tools; Cancer genetic services for TGD patients
Glessner et al. (2012)	Quantitative analyses	General practice	LGBTQ+	Clinical tools; Genetic providers' biases and educational needs; Patient reported barriers to genetic services
Greenwood and Dodelzon (2024)	Review	Cancer	TGD	Cancer genetic services for TGD patients
Heng et al. (2023)	Case report	Cancer	TGD	Cancer genetic services for TGD patients
Hodan et al. (2023)	Professional guideline	Cancer	TGD	Cancer genetic services for TGD patients
Huser et al. (2022)	Quantitative analyses	Cancer; General practice	TGD	Genetic providers' biases and educational needs
Istl et al. (2024)	Quantitative analyses	Cancer	TGD	Cancer genetic services for TGD patients
Jamal et al. (2024)	Commentary	General practice; Research	TGD	Clinical tools
Kamoun et al. (2023)	Commentary	Reproductive	TGD	Conflation of sex, gender, and sex chromosomes
Karmouta et al. (2023)	Case report	Other	TGD	Emerging areas of interest
Li et al. (2018)	Case report	Cancer	TGD	Cancer genetic services for TGD patients
Llorin and Zayhowski (2023)	Commentary	Reproductive	TGD	Conflation of sex, gender, and sex chromosomes
Llorin et al. (2024)	Commentary	Reproductive	TGD	Conflation of sex, gender, and sex chromosomes
Mahon (2023)	Commentary	Cancer	TGD	Clinical tools; Cancer genetic services for TGD patients

(Continues)

TABLE 2 (Continued)

Article	Article type	Specialty focus[a]	Focus: LGBTQ+, LGBQ+, or TGD[b]	Themes extracted
McTiernan et al. (2001)	Quantitative analyses	Cancer	LGBQ+	Cancer care for LGBQ+ individuals
Monseur et al. (2022)	Quantitative analyses	Reproductive	LGBQ+	Reproductive environments
Motiff et al. (2024)	Mixed analyses	General practice	TGD	Clinical tools; Reproductive environments; Genetic providers' biases and educational needs
Mykitiuk and Lee (2015)	Commentary	Reproductive	LGBTQ+	Reproductive environments
Nathan et al. (2019)	Quantitative analyses	General practice	LGBQ+	Genetic providers' biases and educational needs
Palacios-González and Cavaliere (2019)	Commentary	Reproductive	LGBQ+	Reproductive environments
Resta et al. (2023)	Commentary	General practice	LGBTQ+	Clinical tools
Rolf et al. (2021)	Mixed analyses	Cancer	LGBTQ+	Clinical tools; Patient reported barriers to genetic services
Rolle et al. (2022)	Qualitative analyses	Cancer	TGD	Cancer genetic services for TGD patients; Patient reported barriers to genetic services
Roth et al. (2024)	Qualitative analyses	Cancer	TGD	Cancer genetic services for TGD patients; Patient reported barriers to genetic services
Ruderman et al. (2021)	Qualitative analyses	Reproductive	TGD	Clinical tools; Reproductive environments; Genetic providers' biases and educational needs
Sacca et al. (2019)	Case report	Cancer	TGD	Clinical tools; Cancer genetic services for TGD patients
Saunders et al. (2024)	Qualitative analyses	General practice	LGBTQ+	Genetic providers' biases and educational needs
Sehgal (2023)	Review	Other	TGD	Emerging areas of interest
Sheehan et al. (2020)	Mixed analyses	General practice	TGD	Clinical tools; Genetic providers' biases and educational needs
Sieberg et al. (2021)	Case report	Cancer	TGD	Cancer genetic services for TGD patients
Stevens et al. (2023)	Mixed analyses	Reproductive	TGD	Reproductive environments; Conflation of sex, gender, and sex chromosomes; Genetic providers' biases and educational needs
Sutherland et al. (2020)	Review	Cancer	TGD	Cancer genetic services for TGD patients
Theisen and Amarillo (2021)	Review	Laboratory; Research	TGD	Clinical tools; Conflation of sex, gender, and sex chromosomes
Tuite et al. (2020)	Commentary	General practice	TGD	Clinical tools

TABLE 2 (Continued)

Article	Article type	Specialty focus[a]	Focus: LGBTQ+, LGBQ+, or TGD[b]	Themes extracted
Tyrie et al. (2024)	Qualitative analyses	Reproductive	TGD	Reproductive environments; Conflation of sex, gender, and sex chromosomes; Patient reported barriers to genetic services
Valentine et al. (2023)	Mixed analyses	General practice	LGBTQ+	Genetic providers' biases and educational needs; Patient reported barriers to genetic services
VandenLangenberg et al. (2012)	Qualitative analyses	General practice	LGBQ+	Clinical tools; Patient reported barriers to genetic services
von Vaupel-Klein and Walsh (2021)	Review	Cancer; General practice; Other	TGD	Clinical tools; Cancer genetic services for TGD patients; Emerging areas of interest
Zayhowski et al. (2019)	Qualitative analyses	Cancer	TGD	Clinical tools; Cancer genetic services for TGD patients; Genetic providers' biases and educational needs

[a]Some articles had more than one specialty focus. We have categorized the focuses in alphabetical order, with the exception of "Other."
[b]LGBTQ+ (Lesbian, Gay, Bisexual, Transgender, Queer/Questioning, and/or have a sexual orientations and/or gender identity beyond cisheteronormative conceptions), LGBQ+ (Lesbian, Gay, Bisexual, Queer/Questioning, and/or have a sexual orientation that falls outside of heteronormative ideals), and TGD (transgender and gender-diverse; those whose gender identity does not align with cisnormative conceptions).

accurately represents diverse identities, enhancing comfort and trust among LGBTQ+ patients during genetic counseling (Motiff et al., 2024; Sheehan et al., 2020; Tuite et al., 2020). Bennett et al. (2022) published a practice update to pedigree nomenclature, emphasizing the importance of clarified symbols and language to distinguish sex assigned at birth and gender identity, and advocating for a "gender-first" approach to ensure safe and inclusive clinical environments for TGD individuals.

Other tools, such as medical records systems and visual aids, were also identified as barriers to equitable care in clinical settings. Several articles noted that electronic health record systems and intake and genetic testing forms lack inclusive collection of demographic measures related to sex, gender identity/modality, and sexual orientation, significantly hindering the delivery of appropriate, sensitive, and tailored healthcare (Daly et al., 2023; Jamal et al., 2024; Rolf et al., 2021; VandenLangenberg et al., 2012; von Vaupel-Klein & Walsh, 2021; Zayhowski et al., 2019). For instance, Giblin et al. (2023), in a chapter on transgender-inclusive cancer genetic counseling, noted that inadequate medical record systems and sex- and gender-related data collection can lead to clinical confusion, causing TGD patients to be overlooked in routine cancer screenings and preventive services. Bland et al. (2024) illustrated through case analyses from a clinical genomics study how non-inclusive sex- and gender-related survey measures and data models result in inappropriate care recommendations and misgendering for TGD patients. Additionally, two articles examining genetic counselors' experiences demonstrated a failure to represent LGBTQ+ family structures and identities (e.g., same-sex couples, TGD individuals) in genetic counseling visual aids (Motiff et al., 2024; Ruderman et al., 2021).

3.2.1.2 | *Reproductive environments*

Several articles discussed challenges in and evolving strategies for providing inclusive reproductive genetic services for LGBTQ+ patients (Baecher-Lind et al., 2023; Berro & Zayhowski, 2023; Monseur et al., 2022; Mykitiuk & Lee, 2015; Ruderman et al., 2021; Stevens et al., 2023; Tyrie et al., 2024). Ruderman et al. (2021) investigated genetic counselors' experiences with TGD individuals in reproductive settings, finding that interactions with TGD patients prompted genetic counselors to adopt more inclusive language and resources. Participants highlighted TGD individuals' discomfort in prenatal settings, reports of TGD discrimination, and overall feelings of being inadequately prepared to support TGD patients effectively. Monseur et al. (2022) examined reproductive outcomes for single and coupled intended fathers using assisted reproductive technology and found that egg donation and gestational carriers result in successful live births for this population of parents. In a commentary on promoting gender-inclusive learning in medical education, particularly in reproductive environments, Baecher-Lind et al. (2023) identified three common challenges for educators: inclusive language, anatomy education, and reproductive genetics and genetic counseling. The authors suggested that offering disclaimers about gendered terminology, along with using terms such as parentage, parental lines,

and naming specific gametes (sperm and eggs) instead of male/female or maternal/paternal can enhance inclusivity. Similarly, Motiff et al. (2024) explored inclusive language use among genetic counselors, finding that prenatal counselors were more likely to use terms like "sperm" or "egg" rather than referring to gametes as being from "mom" or "dad." Most genetic counselors, regardless of care specialty, preferred gender-neutral terms such as "partner," "parents," and "siblings" over gendered alternatives. However, only one-third of genetic counselors in this study were comfortable using gender-inclusive language, noting that they often used gendered language for clarity and to promote shared language.

The complex interplay between reproductive choices, genetic testing, and ethical considerations was illustrated by several articles. A recent study on genetic counselor experiences disclosing sex chromosome aneuploidies identified through cell-free DNA screening revealed examples of parental questions about the implications of sex chromosome aneuploidies on sex, gender, and sexual orientation (Burzynski et al., 2024). Atkins (2008) employed an autoethnographic approach to examine the decision-making processes of two lesbian mothers regarding genetic testing during pregnancy. The study demonstrated how contextual factors, such as intersectional identities (including LGBTQ+, disabled, and racial/ethnic identities), psychological investment in the fetus, gestational age, clinicians' guidance, and prior unsuccessful embryo implantations influenced their decision to undergo amniocentesis in one pregnancy but not in another. Cavaliere and Palacios-González (2018) presented an ethical argument that lesbian couples should have access to mitochondrial replacement techniques to enable shared genetic parenthood, focusing on reproductive freedom. In response, Baylis (2018) argued against access for this purpose based on the lack of a right to biological parenthood, while Palacios-González and Cavaliere (2019) clarified their position, countering Baylis and advocating for access.

3.2.1.3 | *Conflation of sex, gender, and sex chromosomes*

Several articles explicitly highlighted concerns about conflating sex, gender, and sex chromosomes, specifically in the context of cell-free prenatal DNA screening (Berro & Zayhowski, 2023; Burzynski et al., 2024; Kamoun et al., 2023; Llorin et al., 2024; Llorin & Zayhowski, 2023; Stevens et al., 2023; Tyrie et al., 2024). These discussions suggested that using such screenings for fetal sex prediction may reinforce rigid gender binary norms by incorrectly implying that sex chromosomes determine sex (as sex is multivariable and influenced by a number of factors beyond sex chromosomes) and that sex is synonymous with gender. These articles included commentaries written by healthcare professionals (Berro & Zayhowski, 2023; Kamoun et al., 2023; Llorin et al., 2024; Llorin & Zayhowski, 2023), studies of genetic counselor perspectives (Burzynski et al., 2024; Stevens et al., 2023), and a study of TGD community perspectives (Tyrie et al., 2024). In a commentary (Dusic et al., 2024) and a review (Theisen & Amarillo, 2021), healthcare professionals highlighted challenges related to evaluating sex chromosomes and the lack of EMR documentation for sex and gender information, which can lead to discrepancies among sex, gender, and sex chromosomes, causing potential delays in care for TGD individuals. Moreover, Dusic et al. (2024) critiqued sex checks that compare reported sex to chromosomal data for reinforcing the discriminatory and inaccurate conception that sex is a single binary, fixed, and innate characteristic, and argued that sex checks create unsafe environments by potentially "outing" TGD and intersex individuals/individuals with VSC in clinical settings.

3.2.1.4 | *Cancer care for LGBQ+ individuals*

The scoping review revealed a limited number of articles addressing cancer care and genetic services inequities for patients who are LGBQ+ (Lesbian, Gay, Bisexual, Queer/Questioning sexual orientations, and/or who have a sexual orientation beyond heteronormative conceptions). Burnett et al. (1999) and Durfy et al. (1999) specifically examined motivators for cancer screening and genetic testing among lesbian women, such as breast cancer worry and perceived risk, and McTiernan et al. (2001) investigated breast cancer risk estimates using the Gail and Claus models for various groups, including lesbians. Eckhert et al. (2023) highlighted that breast cancer patients from LGBTQ+ groups faced diagnostic delays and had higher recurrence rates compared with cisgender heterosexual patients, despite showing no significant differences in screening, genetic testing, or initial treatment timelines, suggesting systemic care disparities.

3.2.1.5 | *Cancer genetic services for TGD patients*

Many of the included articles focused on TGD patients' needs in cancer genetics (Antony et al., 2024; Berro et al., 2020; Coad et al., 2021; Colebunders et al., 2014; Corman et al., 2016; Cortina, 2023; Cortina et al., 2024; Daly et al., 2023; Eckhert et al., 2023; Friedman & Papagiannis, 2024; Giblin et al., 2023; Greenwood & Dodelzon, 2024; Heng et al., 2023; Hodan et al., 2023; Huser et al., 2022; Istl et al., 2024; Li et al., 2018; Mahon, 2023; Rolle et al., 2022; Roth et al., 2024; Sacca et al., 2019; Sieberg et al., 2021; Sutherland et al., 2020; von Vaupel-Klein & Walsh, 2021; Zayhowski et al., 2019). Shared paper topics included (1) the impact of gender-affirming care, such as surgery, on cancer risk assessment, (2) the limited existing data regarding impact of gender-affirming hormone therapies on cancer risk, and (3) the imperative for organ inventories and de-gendered language as it relates to organs and hormones. Several clinical case reports and series focused on how aspects of gender-affirming care intersect with cancer and genetic care (Colebunders et al., 2014; Corman et al., 2016; Friedman & Papagiannis, 2024; Heng et al., 2023; Li et al., 2018; Sacca et al., 2019; Sieberg et al., 2021). Berro et al. (2020) and Zayhowski et al. (2019) demonstrated that cancer genetic counselors have substantial knowledge gaps regarding – and lack of experience with – TGD patients. For example, Berro et al. (2020) identified lack of consensus on breast cancer screening recommendations based on estrogen therapy, as well as on genetic testing of minors prior to initiating hormone therapy. Cortina's (2023) commentary stated that breast and ovarian cancer risk assessment is imperative to informed surgical decision-making (e.g., extent of resection) prior to gender-affirming mastectomies. Furthermore, Cortina et al. (2024) piloted an educational intervention with TGD individuals seeking

gender-affirming mastectomies, finding that over half of the cohort had an elevated breast cancer risk, with most opting for a cancer risk-reducing mastectomy after risk identification.

Roth et al. (2024) and Rolle et al. (2022) used qualitative research to explore the experiences of TGD individuals who have heightened genetic risk for cancer and have seen cancer genetic counselors, respectively. Their findings evidenced that previvorship influences gender expression and exploration, revealing that risk-reducing mastectomies can be simultaneously gender-affirming and traumatic. Several articles discussed how insurance coverage inequities for gender-affirming care versus cancer-related services drive some TGD individuals to seek a hereditary cancer diagnosis to access coverage for medically necessary gender-affirming procedures, such as mastectomy, hysterectomy, and/or oophorectomy (Rolle et al., 2022; Roth et al., 2024; Sacca et al., 2019; Zayhowski et al., 2019). Therefore, the potential to uncover a hereditary cancer predisposition may not be the primary focus for some patients when it comes to accessing cancer genetic testing; in these cases, ensuring fully informed consent regarding the implications of such testing is paramount. Sutherland et al. (2020) conducted a literature review and used illustrative case examples to emphasize gaps in clinical knowledge for targeted cancer risk assessment specifically for adolescent TGD individuals, a population discussed in other cancer-related articles addressing both adults and adolescents (Berro et al., 2020; Mahon, 2023; Sacca et al., 2019; Zayhowski et al., 2019). Hodan et al. (2023) and Daly et al. (2023) offered provider recommendations from professional organizations for cancer screening of TGD individuals related to Lynch syndrome and breast, ovarian, and pancreatic cancer, respectively. Greenwood and Dodelzon (2024) reviewed screening for *BRCA* mutations and discussed screening for TGD individuals. These resources focused on individualizing cancer screening based on organs present and history of medical gender-affirming therapies, such as surgery and hormones. Collectively, these articles underscored the limitations of current healthcare systems in addressing the needs of TGD patients and emphasized the need for evidence-based, inclusive, and personalized genetic care.

3.2.1.6 | *Emerging areas of interest*

Several articles focused on other genetic specialties. Bayram et al. (2024) was the sole study in neurogenetics, examining how sexual orientation and gender influence attitudes toward research and genetic testing among individuals with Parkinson's disease, finding that a higher percentage of LGBTQ+ individuals were concerned about transportation and researchers not respecting their beliefs. Karmouta et al.'s (2023) case report addressing ophthalmic genetics examined familial exudative vitreoretinopathy in the context of a TGD teenager, where knowledge of the patient's gender modality was crucial to understanding the intersection of their genetic carrier status of an *LRP5* mutation, bone health, and their gender-affirming care pathway to ensure comprehensive care. von Vaupel-Klein and Walsh (2021) provided an overview of several topics related to TGD individuals in genetic care and were the only articles to specifically examine the intersection of cardiac genetics with gender-affirming care, highlighting the impact of hormone therapies on cardiovascular risk. Sehgal (2023) reviewed pharmacogenomic considerations related to gender-affirming medications, describing a lack of research into pharmacogenetic interactions with hormone medications used for medical gender affirmation.

3.2.2 | Genetic providers' biases and educational needs

This theme included studies that examined genetic counselors' experiences and attitudes toward LGBTQ+ patients, as well as genetic counselor-reported educational needs (Berro et al., 2020; Burzynski et al., 2024; Ernst et al., 2023; Glessner et al., 2012; Huser et al., 2022; Motiff et al., 2024; Nathan et al., 2019; Ruderman et al., 2021; Saunders et al., 2024; Sheehan et al., 2020; Valentine et al., 2023; Zayhowski et al., 2019). Ruderman et al. (2021), Berro et al. (2020), and Zayhowski et al. (2019) demonstrated a lack of specific training for working with TGD patients in reproductive and cancer settings. Motiff et al. (2024) found that only one-third felt comfortable using gender-inclusive language and just over half of those did so regularly; many did not share or ask for pronouns, indicating a need for more training. Valentine et al. (2023) supported this finding, showing that while most genetic counselors felt comfortable working with LGBTQ+ patients, many patients were not prompted to disclose their identities, exacerbating clinical care inequities and hindering the delivery of personalized, informed care. Nathan et al. (2019) revealed that 60.2% of genetic counselors and student participants had implicit preferences for heterosexual individuals; however, most supported addressing LGBTQ+ health-related needs, suggesting that further curricula in this area may help close bias-related gaps. Multiple studies showed that genetic counselors often lack education on and exposure to LGBTQ+ needs, particularly regarding gender inclusivity and TGD healthcare needs (Berro et al., 2020; Burzynski et al., 2024; Glessner et al., 2012; Motiff et al., 2024; Nathan et al., 2019; Ruderman et al., 2021; Sheehan et al., 2020; Stevens et al., 2023; Valentine et al., 2023; Zayhowski et al., 2019). Together, these articles suggested a need to develop and distribute tailored genetic counselor education regarding this population.

Consistent with these findings, several articles highlighted the necessity for genetic providers to receive more comprehensive education on LGBTQ+ care, stressing the importance of community involvement in creating educational initiatives and tools. For several articles, education was a foundational aim (Berro et al., 2020; Ernst et al., 2023; Huser et al., 2022; Saunders et al., 2024). Saunders et al.'s (2024) qualitative study of genetic counselor educators who teach on LGBTQ+ topics underscored the need for developing and integrating LGBTQ+-specific competencies into genetic counseling graduate programs to address health disparities and prepare students to provide inclusive care. The creation of educational modules focused on gender-affirming care has been identified as a promising strategy to enhance the competencies of genetic counselors (Ernst et al., 2023; Huser et al., 2022).

3.2.3 | Patient-reported barriers to genetic services

This theme encompassed studies examining LGBTQ+ patients' experiences and perceptions relating to genetic services (Angelo et al., 2022; Antony et al., 2024; Atkins, 2008; Barnes et al., 2020; Bayram et al., 2024; Burnett et al., 1999; Chen et al., 2017; Durfy et al., 1999; Glessner et al., 2012; Rolf et al., 2021; Rolle et al., 2022; Roth et al., 2024; Tyrie et al., 2024; Valentine et al., 2023; VandenLangenberg et al., 2012). Angelo et al. (2022) conducted a study on medical distrust within diverse communities in the context of genomic medicine, identifying that TGD individuals experience notably high levels of medical distrust. Rolf et al. (2021) and Barnes et al. (2020) specifically examined challenges related to family history collection for LGBTQ+ individuals, while others emphasized communication barriers that arise when discussing genetic risk within families (Barnes et al., 2020; Rolle et al., 2022; Roth et al., 2024). These studies suggested that strained familial relationships (common among LGBTQ+ individuals due to familial identity rejection) can limit knowledge of family health history and impede communication of genetic testing results to at-risk relatives. Additionally, several studies explored the impact of overt discrimination or anticipation of discrimination on participants (Rolle et al., 2022; Roth et al., 2024; Tyrie et al., 2024; Valentine et al., 2023). In one study, TGD patients reported anticipatory anxiety about potential physical exams during cancer genetic counseling appointments, stemming from prior experiences of medical discrimination during physical exams (Rolle et al., 2022). The authors suggested that, to improve appointment adherence, genetic counselors should proactively inform patients if an exam is required and clarify what to expect. Glessner et al. (2012), VandenLangenberg et al. (2012), and Valentine et al. (2023) found that while counselors often believe they are promoting a supportive environment, LGBTQ+ patients may hesitate to disclose their sexual orientation or gender identity due to concerns about discrimination or discomfort, underscoring the need for enhanced communication strategies and counselor training.

4 | DISCUSSION

This scoping review explored the landscape of LGBTQ+ care in clinical genetics, exemplifying the unique challenges and needs of LGBTQ+ individuals within this domain. The analysis included 65 articles that collectively shed light on the experiences, perceptions, and care practices related to genetic services among LGBTQ+ populations.

4.1 | Addressing insufficient data collection practices

The lack of consistent, comprehensive data collection on sex, gender, and sexual orientation significantly limits the capacity of genetic counselors and healthcare systems to deliver individualized, informed care to LGBTQ+ patients. Inconsistent data practices not only obscure health risks but also impair accurate risk assessment, especially for TGD patients who have undergone gender-affirming medical interventions (Berro et al., 2020; Berro & Zayhowski, 2023; Bland et al., 2024; Jamal et al., 2024; Rolle et al., 2022; Sutherland et al., 2020; Theisen & Amarillo, 2021; Zayhowski et al., 2019). Many current EHRs do not include fields for critical information, such as sex assigned at birth, gender modality, and organ inventories, and/or these variables are inconsistently documented leading to missed opportunities to analyze long-term health outcomes for LGBTQ+ populations (Albert & Delano, 2022; Bland et al., 2024; Jamal et al., 2024; Lau et al., 2020; Sokkary et al., 2021). Improving and standardizing EHR variable collection, alongside regularly updating patient records, would enable more precise cancer risk assessments, for example (Bland et al., 2024). This lack of accurate and inclusive data also underscores the need for standardized testing requisitions and protocols across laboratories and clinics to improve the quality of care and enhance the experience of LGBTQ+ patients (Bland et al., 2024; Dusic et al., 2024; Jamal et al., 2024; Theisen & Amarillo, 2021; Zayhowski et al., 2025).

The deficit of inclusive data practices and consistent terminology creates significant barriers to accurate cancer risk assessment and access to tailored clinical genetic services (Barnes et al., 2020; Berro et al., 2020; Bland et al., 2024; Jamal et al., 2024). Hormone therapy and gender-affirming surgeries can influence cancer risk profiles for TGD individuals; however, insufficient data and limited understanding of these interventions lead to incomplete risk assessments and care gaps (Berro et al., 2020; Daly et al., 2023; Giblin et al., 2023; Hodan et al., 2023; Mahon, 2023; Sacca et al., 2019; Sutherland et al., 2020; von Vaupel-Klein & Walsh, 2021; Zayhowski et al., 2019). There is an urgent need for further research to clarify how gender-affirming therapies affect cancer risk and to incorporate more precise measures such as organ inventories and hormonal milieus into risk assessment models. To effectively address these barriers, genetic counselors require specialized training in TGD cancer care, coupled with the integration of inclusive gender- and sex-related variables in health records and clinical and laboratory forms.

Individualized genetic risk assessments and recommendations for cancer risk reduction should consider factors such as age, family history, genetic status, surgical history, and duration of hormone therapy. This tailored approach must prioritize specific organs most at risk based on the patient's organ inventory and hormonal milieu, particularly in light of the impact of surgeries and hormone therapies on cancer risk assessments (Bland et al., 2024; Cortina, 2023; Cortina et al., 2024; Daly et al., 2023; Roth et al., 2024; Sutherland et al., 2020; von Vaupel-Klein & Walsh, 2021; Zayhowski et al., 2019). Importantly, TGD patients should have access to cancer genetic testing prior to gender-affirming mastectomy to inform surgical decisions (Berro & Zayhowski, 2023; Cortina, 2023; Cortina et al., 2024). However, they should not be denied gender-affirming care, such as hormone therapy, based solely on genetic results (Berro & Zayhowski, 2023). Ultimately, improving data collection practices

is essential to enhance genetic risk assessment accuracy and inclusivity and to ensure equitable healthcare for LGBTQ+ individuals.

Given the rise in anti-LGBTQ+ policies in the United States (American Civil Liberties Union, 2024; Kinney et al., 2022; Zayhowski et al., 2025), disclosing sex-, gender-, or sexual orientation-related information can be unsafe for patients, making its documentation in medical records precarious. Patients may refrain from disclosure for fear of discrimination (by the provider or by other providers who see documentation in the medical record) or out of concerns for privacy of their LGBTQ+ status (Friley & Venetis, 2021; Kamen et al., 2022; Maragh-Bass et al., 2017; Sequeira et al., 2020; Thompson, 2016). For example, Vanderbilt University Medical Center recently faced backlash for releasing full TGD patient records to the Tennessee Attorney General amid a Medicaid fraud investigation, raising serious concerns about patient privacy (American Medical Association, 2023; Kohli, 2023; Zayhowski et al., 2025). This incident highlights LGBTQ+ community vulnerability and the importance of documentation practices that protect patient safety by ensuring that sensitive data are used only for its intended purpose (Xu & Zhang, 2019). While the Health Insurance Portability and Accountability Act (HIPAA) generally safeguards patient privacy, its protections are limited when faced with government interception (Clayton et al., 2024; Zayhowski et al., 2025). In the current United States climate, where laws increasingly limit or even criminalize the actions of LGBTQ+ individuals and restrict their healthcare access, risks associated with the use of their data are heightened (Clayton et al., 2024; Kraft & Mittendorf, 2024; Zayhowski et al., 2025). The principle of "do no harm" requires protective policies at institutional and governmental levels. To mitigate privacy risks, healthcare institutions should implement explicit consent protocols, robust data encryption, and strict policies against unnecessary disclosures, as well as consider data segmentation options (Clayton et al., 2024; Kraft & Mittendorf, 2024; Zayhowski et al., 2025). Training healthcare providers on the ethical management of sensitive information is crucial to mitigate misuse, maintain confidentiality and promote patient trust, especially in today's hostile sociopolitical climate.

4.2 | Cultivating inclusive language in genetic services

Non-inclusive language creates adverse environments and deters LGBTQ+ patients from full genetic appointment participation (Baecher-Lind et al., 2023; Motiff et al., 2024; Rolle et al., 2022; Roth et al., 2024). Language that reflects cisheteronormative assumptions can alienate patients and reduce the trust necessary for effective counseling (Berro & Zayhowski, 2023; Motiff et al., 2024; Rolle et al., 2022). For instance, non-inclusive terminology may prevent patients from disclosing relevant information about their identities or family structures (Motiff et al., 2024; Rolf et al., 2021; Roth et al., 2024; Ruderman et al., 2021). Recent updates to pedigree nomenclature guidelines represent progress toward gender inclusivity (Bennett et al., 2022), and this inclusivity must be integrated into other aspects of genetic clinical sessions. Clinicians should prioritize the use of inclusive language, demonstrate humility in acknowledging errors, and consistently recognize and affirm patients' gender identities (von Vaupel-Klein & Walsh, 2021). It is essential for providers to offer counseling that is both LGBTQ+ inclusive and accessible to those with limited education, in resource-scarce settings, or facing language barriers, underscoring the need to develop tools and resources that can effectively serve diverse patient populations. Inclusive language training should be integral to medical education; training should include guidance on pronoun use and correct gender modality terms, and challenge preconceived assumptions about family structures and reproductive contexts (Baecher-Lind et al., 2023; Motiff et al., 2024; Ruderman et al., 2021). Clinician humility—demonstrating openness to correction and respectful dialogue when mistakes are made—is critical for establishing trust. Combining the strategies outlined above could help alleviate patients' anticipatory anxiety, improve communication, and enhance overall satisfaction with care (Barnes et al., 2020; Rolle et al., 2022; Roth et al., 2024).

Language surrounding gender and sexual orientation has historically been misused in healthcare settings, reinforcing stereotypes and stigmatizing LGBTQ+ patients, especially when rigid definitions or outdated terminology are used. A major ethical concern lies in the conflation of sex, gender, and sex chromosomes, particularly in processes like cell-free prenatal DNA screening, which can perpetuate harmful gender binaries and reinforce stereotypes within clinical care and broader society (Burzynski et al., 2024; Kamoun et al., 2023; Llorin et al., 2024; Llorin & Zayhowski, 2023; Ruderman et al., 2021; Stevens et al., 2023; Tyrie et al., 2024). Traditional laboratory practices, such as "sex checks" that compare reported sex with chromosomal data, create unsafe environments for TGD individuals (Dusic et al., 2024; Theisen & Amarillo, 2021). The National Society of Genetic Counselors recently issued a position statement underscoring the harms of conflating sex and gender with chromosomal data and calling for more inclusive practices (National Society of Genetic Counselors, 2024). The recurrent conflation of sex- and gender-related variables identified across numerous articles underscores the need for a standardized, nuanced approach that respects individual identities and minimizes clinical ambiguity (Burzynski et al., 2024; Jamal et al., 2024; Llorin et al., 2024; Motiff et al., 2024; Stevens et al., 2023; Theisen & Amarillo, 2021; Tyrie et al., 2024). Guiding organizations in genetics must establish clear guidelines that delineate sex- and gender-related concepts and provide structured guidance on their appropriate discussion and collection in clinical settings. Clinicians should proactively use gender-inclusive terminology and clearly communicate the intended use of sex- and gender-related information in counseling and testing (Dusic et al., 2024; Jamal et al., 2024; Llorin et al., 2024; Mahon, 2023; Motiff et al., 2024). Collaboration with LGBTQ+ advocacy organizations or other community engagement could further align clinical language and protocols with best practices, minimizing potential for harm through stigmatizing or inaccurate language (Ernst et al., 2023;

Huser et al., 2022; Key et al., 2019; Long et al., 2022; Zayhowski et al., 2024).

4.3 | Combating bias and discrimination in care delivery

Overt discrimination and implicit bias in genetic services perpetuate health inequities and can severely undermine open dialogue (Nathan et al., 2019). Medical distrust can discourage LGBTQ+ patients from sharing crucial personal or familial information necessary for accurate risk assessment (Angelo et al., 2022; Antony et al., 2024; Berro & Zayhowski, 2023; Nathan et al., 2019; Valentine et al., 2023). Research indicates that many LGBTQ+ individuals feel unsafe disclosing their identities to healthcare providers, resulting in incomplete assessments and compromised health outcomes (Glessner et al., 2012; Jamieson et al., 2020; Kamen et al., 2015; Valentine et al., 2023; VandenLangenberg et al., 2012; Whitehead et al., 2016). Genetic counselors must also be attuned to the challenges of gathering family histories from LGBTQ+ patients, as disruptions in familial relationships are common and can impede complete risk assessments (Giblin et al., 2023; Rolf et al., 2021; Rolle et al., 2022). For TGD patients, proactive communication about appointment expectations—including whether physical examinations will occur—can alleviate anticipatory anxiety, enhancing both adherence and patient comfort (Mahon, 2023; Rolle et al., 2022; von Vaupel-Klein & Walsh, 2021). Genetic counseling training materials should incorporate information on the sensitivity of family history collection, family communication, and physical exams in this population.

Genetic counselors have reported feeling unprepared to meet the needs of LGBTQ+ individuals, highlighting an urgent need for expanded training on the complexities of LGBTQ+ health (Berro et al., 2020; Motiff et al., 2024; Saunders et al., 2024; Valentine et al., 2023; Zayhowski et al., 2019). Although some training programs include LGBTQ+ topics, many genetic counselors still express uncertainty about providing inclusive care, as current curricula often lack the depth needed to fully equip them to serve these populations (Ernst et al., 2023; Motiff et al., 2024). As such, there is a need for continuous evaluation and enhancement of educational resources, supplemented by community-led initiatives to create a more intentional and responsive genetic healthcare environment.

Incorporating a restorative justice framework within genetic services could provide substantial benefits (Berro & Zayhowski, 2023; Long et al., 2022; Zayhowski et al., 2024). Rooted in healing, accountability, and community involvement, this approach invites open dialogue with LGBTQ+ individuals and community representatives, allowing them to voice their experiences and contribute insights to shape practice. Community-informed education models can build and sustain trust, offering a foundation for advocacy and mutual learning (Berro & Zayhowski, 2023; Ernst et al., 2023; Huser et al., 2022; Long et al., 2022). By partnering with LGBTQ+ advocacy organizations, genetic practices can integrate community perspectives into ethical guidelines, policies, and research protocols, promoting trauma-informed care that centers on LGBTQ+ patients' needs.

4.4 | Research directions

This scoping review highlights a significant increase in LGBTQ+-related research, a trend that is similarly observed across various other fields (An & Batra, 2022), reflecting growing recognition of the importance of inclusivity and representation in academic inquiry. However, despite this progress, the reviewed studies revealed substantial gaps in the existing literature concerning culturally responsive genetic care for LGBTQ+ individuals. A notable limitation identified in this review is the predominance of commentaries, qualitative studies, and case reports. While these article types provide valuable insights into LGBTQ+ individuals' and healthcare providers' experiences and perspectives, they often lack the generalizability of quantitative research or the clinical applicability of implementation studies. The reliance on these articles makes it challenging to draw broad conclusions or establish definitive trends regarding the provision of inclusive genetic services.

Sex-, gender-, and sexual orientation-related variables are often excluded or inadequately represented in large epidemiological datasets, underscoring the need for improved data collection on these factors (Bland et al., 2024; Jamal et al., 2024; Kamen et al., 2022; Maragh-Bass et al., 2017; Streed Jr et al., 2020). Additionally, shifting societal attitudes, evolving terminology, discrimination in research funding, and mistrust within the LGBTQ+ community further hinder large-scale research efforts (Bland et al., 2024; Hammack-Aviran et al., 2022; Rajkovic et al., 2022; Zayhowski et al., 2025). Future research should prioritize the development of large-scale quantitative and implementation studies to evaluate the inclusivity of current genetic services and the effectiveness of interventions to improve genetic services delivery for LGBTQ+ individuals. Such studies can yield robust data to inform best practices and facilitate evidence-based improvements in clinical settings.

Another critical concern is the demographic limitations of the studies included in this review. Many investigations and commentaries have focused primarily on practitioners' perspectives rather than the voices of LGBTQ+ patients themselves. This practitioner-centric approach overlooks the experiences and needs of LGBTQ+ individuals seeking genetic services. By failing to involve LGBTQ+ individuals as partners in research design, implementation, and policy development, healthcare practices perpetuate exclusion and reinforce systemic inequities (Key et al., 2019; Zayhowski et al., 2024). Neglecting to employ community-engaged approaches throughout the research process can significantly harm marginalized communities by leading to misunderstandings, misrepresenting their needs, and producing interventions that fail to address real challenges. Engaging LGBTQ+ individuals in community-based research is essential to ensure their voices are central to the development of effective genetic practices that align with community priorities (Berro & Zayhowski, 2023; Key et al., 2019; Long et al., 2022; Zayhowski et al., 2024).

Recognizing and reflecting on researcher positionality is crucial in the design and conduct of studies focused on LGBTQ+ populations (Topp et al., 2021; Wilson et al., 2022). Researcher members of the LGBTQ+ community bring valuable insights and lived experiences that enhance the relevance and sensitivity of research methodologies. Their involvement promotes research trust among participants and helps ensure that the needs and perspectives of the community are adequately represented in research design and analysis. To mitigate the risks of misalignment and to promote meaningful engagement, it is invaluable to prioritize the perspectives of patients themselves by collaborating with patient partners throughout study design and implementation. This strategy can lead to more relevant and impactful research outcomes that effectively address the challenges faced by marginalized groups (Bukamal, 2022; Topp et al., 2021; Zayhowski et al., 2024).

Research focused on the experiences of LGBQ+ individuals remains critically limited. This oversight poses significant challenges to understanding the experiences and healthcare requirements of LGBQ+ individuals seeking genetic services. While there has been some exploration of TGD individuals, the volume of studies addressing their needs remains insufficient. Current literature overwhelmingly concentrates on cancer genetics, leaving significant gaps in other specialties such as pediatrics, broader adult genetics, and prenatal care. Moreover, much of the existing cancer literature specifically discusses the dearth of long-term data, and many articles comment on the deficiency of information regarding the impact of gender-affirming care, such as surgeries and hormone therapy, on cancer risk (Berro et al., 2020; Coad et al., 2021; Daly et al., 2023; Hodan et al., 2023; Sutherland et al., 2020; von Vaupel-Klein & Walsh, 2021; Zayhowski et al., 2019). To effectively gather necessary data, it is crucial to establish policies and practices that allow individuals to safely access medical services without fear of discrimination or unsafe data-sharing practices, creating an environment where they can safely disclose aspects of their identities. Only then can improvements to electronic health record systems and data collection methodologies be meaningfully achieved (Bland et al., 2024; Jamal et al., 2024).

4.5 | Limitations

This scoping review is subject to several limitations that may affect the comprehensiveness and applicability of its findings. One significant limitation is the potential for publication bias, as inclusion criteria were restricted to English-language publications and databases primarily indexing Western literature. This approach may exclude valuable insights and practices from non-English-speaking regions and diverse cultural contexts, limiting our understanding of LGBTQ+ healthcare experiences globally. Furthermore, the geographic bias inherent in our search strategy could restrict the representation of LGBTQ+ healthcare experiences from regions where disparities and cultural nuances differ significantly from those in Western countries. As a result, the review may not fully capture the breadth of insights necessary for promoting global LGBTQ+ inclusivity in genetics healthcare. Additionally, the dynamic nature of LGBTQ+ terminology presents another limitation. Our search may have missed relevant literature that employs terms not included in our search criteria, leading to an incomplete overview of the current state of LGBTQ+ inclusivity in genetic services and healthcare settings. This limitation highlights the necessity for ongoing adaptations in language and understanding within the field to ensure all relevant perspectives are considered. Additionally, the search strategy was limited by database indexing, specifically the inclusion and comprehensiveness of titles, subject headings, and abstracts, leading to the potential omission of relevant articles that did not contain search terminology. In this scoping review, we included non-original research, such as commentaries, due to the limited availability of evidence on the topic. Additionally, bias assessments were not conducted for the included studies. Lastly, it is important to note that other types of articles, such as book chapters on these topics, which were not indexed in the databases we searched, would not have been retrieved.

5 | CONCLUSIONS

This scoping review underscores the pressing need for more comprehensive and inclusive approaches to genetic healthcare for LGBTQ+ individuals. While existing literature provides valuable insights, it is limited by a reliance on commentaries and case studies and a lack of patient perspectives. To advance the field, future research must prioritize a diversity in methodologies and center the voices of LGBTQ+ patients. In practice, an essential area of focus should be the education of genetic providers, as well as the evaluation of training initiatives aimed at enhancing providers' competency in inclusive care practices. Improved documentation and data practices related to sex- and gender-related variables and organ inventory are also necessary. A standardized approach to these data fields, informed by a commitment to respect, privacy, and accuracy, would help reduce the risks of inappropriate or discriminatory practices, such as conflating sex with gender, applying sex-related criteria inaccurately, or disclosure to unsafe parties. By embracing a more holistic research agenda that incorporates diverse populations and patient-centered perspectives, we can develop evidence-based practices that address the needs of LGBTQ+ individuals in genetic services. Ultimately, this commitment to inclusivity will not only improve the quality of care but also promote a healthcare environment where LGBTQ+ individuals feel understood, respected, and empowered to make informed decisions about their health.

AUTHOR CONTRIBUTIONS

Kimberly Zayhowski: Conceptualization; methodology; validation; formal analysis; investigation; writing – original draft; writing – review and editing; visualization; project administration; funding acquisition. Kayla Horowitz: Conceptualization; methodology; validation; formal analysis; investigation; writing – original draft; writing – review and editing; visualization. Molly Bostrom: Methodology; software;

investigation; resources; data curation; writing – review and editing. Kathleen F. Mittendorf: Conceptualization; formal analysis; writing – original draft; writing – review and editing; visualization. Megan Kocher: Methodology; software; investigation; resources; data curation; writing – review and editing. Jehannine (J9) Austin: Conceptualization; writing – review and editing; supervision. Ian M. MacFarlane: Conceptualization; methodology; validation; formal analysis; writing – review and editing; supervision; funding acquisition.

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CONFLICT OF INTEREST STATEMENT

Kimberly Zayhowski, Kayla Horowitz, Molly Bostrom, Megan Kocher, Jehannine (J9) Austin, and Ian M. MacFarlane declare that they have no conflicts of interest. Kathleen F. Mittendorf declares institutional support from GE Healthcare for unrelated work and a financial interest in Lavender Spectrum Health, a small private primary care practice that predominantly serves LGBTQ+ patients.

DATA AVAILABILITY STATEMENT

Data collected for this study are available from the authors upon request.

ETHICS STATEMENT

Human studies and informed consent: No human subjects research was carried out by the authors for this scoping review.

Animal studies: No nonhuman animal studies were carried out by the authors for this scoping review.

ORCID

Kimberly Zayhowski https://orcid.org/0000-0003-1712-6403
Kayla Horowitz https://orcid.org/0000-0002-8319-2188
Molly Bostrom https://orcid.org/0009-0003-3754-6218
Kathleen F. Mittendorf https://orcid.org/0000-0003-1097-9171
Megan Kocher https://orcid.org/0000-0001-8200-5144
Jehannine (J9) Austin https://orcid.org/0000-0003-0338-7055
Ian M. MacFarlane https://orcid.org/0000-0002-0896-1952

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SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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