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Gender on the Spectrum: Prevalence of Gender Diversity in Autism Spectrum Disorder—A Systematic Review and Meta-Analysis

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Abstract

Background: The intersection of Autism and Gender Diversity (GD) is an emerging field of research. While prior studies have primarily focused on autistic traits within gender-diverse populations, limited evidence exists on the prevalence and characteristics of GD among individuals with a confirmed Autism Spectrum Disorder (ASD) diagnosis. This study aims to fill this gap through a systematic review and meta-analysis, offering comprehensive insights into the prevalence of GD in autistic individuals and the factors influencing this intersection.

Methods: We conducted a systematic review and meta-analysis following the Preferred Reporting Items for Systematic Reviews and Meta-analyses 2020 guidelines. We searched for studies published between 2013 and 2023 in four databases. Inclusion criteria focused on studies assessing GD in children and adult participants with a formal ASD diagnosis based on *Diagnostic and Statistical Manual of Mental Disorders*, Fifth Edition, criteria. We used random-effects models to estimate prevalence rates, and subgroup analyses explored potential influencing factors, including age, gender assigned at birth, type of GD assessment, ASD diagnosis methodology, and primary objective of the study.

Results: From 7133 identified reports, we deemed 24 studies suitable for the systematic review, with 14 meeting all eligibility criteria for meta-analysis, representing 3894 autistic participants. We estimated the pooled prevalence of GD at 7.37% (95% confidence interval [CI]: 4.45–11.98). Subgroup analyses revealed no significant impact of age, type of GD assessment, or diagnostic methodology of ASD on GD prevalence. Although we found a higher prevalence of GD among individuals assigned female at birth (14.54%) compared with those assigned male at birth (8.15%), the difference was not statistically significant. However, studies explicitly addressing GD reported statistically higher GD rates (13.71%) compared with those with other primary objectives (4.71%). Non-binary identities appeared particularly prevalent, but we did not underexplore it due to variability in GD definitions across studies.

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Conclusion: This study highlights a notable prevalence of GD among autistic individuals, emphasizing the need for inclusive definitions and diagnostic process. Understanding this intersection is crucial for enhancing support services and ensuring equitable access to gender-affirming care for neurodiverse populations.

Keywords: autism spectrum disorder, gender diversity, transgender, non-binary

Community Brief

Why is this topic important?

This study looks at how Autism Spectrum Disorder (ASD) and Gender Diversity (GD) overlap. Autistic and gender-diverse people face unique challenges and social stigmas. Understanding how these two areas intersect can help us create better support systems for those with both. By learning how common GD is among autistic people, we can help health care providers, teachers, and policymakers better support this group.

What is GD?

GD means that people's experiences of gender go beyond just being male or female. It includes individuals who identify as transgender, non-binary, genderqueer, or other gender identities that do not fit traditional binary notions of male and female.

What is the purpose of this article?

The purpose of this article is to find out how many autistic people also identify as gender diverse and to identify factors that influence this prevalence. The authors want to fill gaps in current research using clear criteria for diagnosing ASD and a broad definition of GD. They aim to improve our understanding of how ASD and GD are related.

What did the authors do?

The authors reviewed and analyzed research from January 2013 to March 2023. They found 24 studies that met their criteria and used data from 14 of these studies to estimate how common GD is among autistic people and to understand the factors that might influence this.

What did the authors find?

The authors found that about 7 out of 100 autistic people also identify as gender diverse (7.37%). They are fairly confident that the true number is between 4 and 12 out of 100 (confidence interval between 4.45% and 11.98%). This rate did not seem to change based on age or sex at birth. However, studies that focused on gender identity showed higher rates of gender diversity. There was not a specific type of gender diversity found among autistic people, but non-binary identities were common.

What do the authors recommend?

The authors recommend that future research should continue to explore the intersection of ASD and GD, including non-binary and other diverse gender identities. The authors also emphasize the importance of health care providers being aware of the high rate of GD among autistic people and ensuring that support services are inclusive and tailored to their specific needs.

How will these findings help autistic adults now or in the future?

By showing clearly how common GD is among autistic people, this research highlights the need for inclusive health care services. Recognizing GD in this group can lead to better-targeted health care support and interventions. In the future, these findings can guide policy changes and the development of training programs for health care professionals, ensuring that they are prepared to support autistic individuals who are also gender diverse.

Background

Autism is a part of human diversity, where social interactions often feel like a foreign language and daily routines are a source of comforting predictability. Defined by the American Psychiatric Association, Autism Spectrum Disorder (ASD) is a neurodevelopmental condition that affects how a person perceives and interacts with others and is characterized by difficulties in social communication and interaction, as well as repetitive or restricted interest and behaviors.[1]

Gender Diversity (GD) is a broad concept that encompasses the various ways in which a person's gender identity might differ from the gender they were assigned at birth. This can include individuals who identify as transgender, non-binary, genderqueer, or other gender identities that do not fit in traditional binary notions of male and female. GD acknowledges that gender is a spectrum and that people's experiences and identities can be complex and multifaceted. Current literature exploring the intersection of autism and GD reveals that a notable proportion of autistic people identify themselves as gender diverse.[2] This means that they might experience a gender identity that does not align with societal expectations based on their assigned gender at birth.

In fact, in recent years, only a handful of systematic reviews and meta-analyses have explored this question. In this article, we aim to fill that gap by conducting both a systematic review of the literature, to offer a comprehensive narrative synthesis concerning autism and GD, and a meta-analysis to precise our qualitative conclusions and measure the prevalence of GD in autistic people. Previous reviews have been mainly focusing on medical conditions defined in classical nosography as follows: (1) *gender dysphoria* defined in the *Diagnostic and Statistical Manual of Mental Disorders*, Fifth Edition (DSM-5), as a clinically significant distress or impairment caused by a persistent incongruence between one's sex assigned at birth and one's gender identity; and/or (2) *gender identity disorders* (GID) in International Classification of Diseases (ICD)-10 (1994) characterized by a persistent and intense distress about assigned sex, together with a desire to be (or insistence that one is) of the other sex.[3] The more recent diagnosis of *gender incongruence* from the ICD-11 (published in 2019), characterized by a marked and persistent incongruence between an individual's experienced gender and the assigned sex, was too recent to be found in published literature yet.[4] In this article, we will use the terms "Gender Dysphoric/GID" or "Gender Dysphoria/GID" when referring to studies based on those medical concepts. Contrariwise, we will use the broader and more inclusive term "GD," as defined earlier, when discussing the diversity found in gender identities, beyond medical classifications.

Autism in gender-diverse people

Most of the existing literature focuses on autistic traits within populations diagnosed with Gender Dysphoria/GID. For instance, in a narrative review published in 2016, van der Miesen et al.[2] examined 25 articles (original data studies and case reports as well as letters and expert opinion articles). They studied the rate of intersection between gender dysphoria and autism in eight original data studies in adults and children and found that autism is frequently co-occurring with gender dysphoria, with rate ranging from 3.8% to 21.3%, depending on the studies. In 91 adults with gender dysphoria, Pasterski et al.[5] showed that the prevalence of autistic traits was 5.5%, which is superior to clinical diagnosis of 0.5% to 2% in the general population. Similar findings have been observed in children and adolescents; for example, De Vries et al.[6] identified a 7.8% incidence of ASD diagnoses in a sample of children and adolescents referred to gender clinics, whereas Skagerberg et al.[7] noted that over 54% of gender-diverse children scored within the mild-to-severe range on the Social Responsiveness Scale.[8] This measure identifies social impairment associated with autism, distinguishes ASD from other disorders, and is interpreted as a quantitative measure of autistic features. Other studies, such as the 2016 systematic review by Glidden et al.,[9] have shown that the prevalence of autism in children and adolescents with gender dysphoria is higher than in the general population, but research on adults remains limited.

A more recent meta-analysis by Kallitsounaki and Williams[10] revealed that 11% of individuals with Gender Dysphoria/GID also had an ASD diagnosis. However, most studies emphasize autistic traits within gender-diverse populations, with fewer investigating the prevalence of GD among individuals with confirmed autism diagnoses. In addition, the prevalence of autism in gender-diverse populations has been shown to fluctuate across different age-groups—children, adolescents, and adults—yet the potential impact of age on this relationship remains unexplored.

GD in autistic people

In a few studies, researchers focused specifically on Gender Dysphoria/GID within the autistic population, highlighting other factors worth investigating. For instance, one study utilizing the Gender-Identity/Gender-Dysphoria Questionnaire for Adolescents and Adults[11] found that autistic individuals reported a higher number of gender-dysphoric symptoms compared with neurotypical individuals.[12] In addition, the meta-analysis by Kallitsounaki and Williams[10] demonstrated a significant relationship between autistic traits and Gender Dysphoria/GID feelings in the general population. However, the exact prevalence of Gender Dysphoria/GID within the autistic population remains unknown.

In a study involving 675 autistic adolescents and adults, Dewinter et al.[13] found that approximately 22% of women and 8% of men reported "gender non-conforming feelings." Although the study did not permit statistical comparison, the difference in prevalence between women and men suggests a potential role of assigned gender at birth, raising important questions for further research.

Autism and GD: Expanding the spectrum

Beyond the notions of Gender Dysphoria/GID, autistic people appear to present with a broader spectrum of gender identities, encompassing one's innermost feeling of maleness, femaleness, a blend of both, or neither. This broader spectrum is better encapsulated by the term GD used in this article. We therefore also considered the possibility that a specific type of GD might be more prevalent than others within the autistic population.

Furthermore, much of the international literature uses a broad definition of autism, ranging from autistic traits and psychometric screening measures to self-diagnosed autism, which diverges from the current diagnostic criteria for ASD. In studies involving gender diverse individuals, authors often evaluated autism using the Autism Spectrum Quotient (AQ[14]), a self-report questionnaire comprising 50 items that assess the extent of autistic traits via a 4-point Likert scale. However, while the AQ serves as a valuable tool for measuring autistic traits in the general population, it lacks the capacity to diagnose ASD, particularly within gender diverse populations.[15] According to Mazzoli et al.,[16] autistic traits in individuals with

gender dysphoria may be more of an epiphenomenon than a direct sign of autism, as evidenced by their significant decrease following one year of gender-affirming hormonal therapy. To ascertain whether the intersection of autism and GD is a genuine phenomenon rather than an epiphenomenon, it is imperative to evaluate the prevalence of GD within a population with a robust diagnosis of ASD.

Finally, we believe it is important to explain why this research is significant to us as the primary coauthors. We work daily in welcoming and supporting autistic individuals in a specialized university-hospital unit. Through this work, we have encountered many gender-diverse individuals. The specific challenges they face prompted us to explore this topic. With this work we aim to better understand the situation of this dual diversity to ultimately improve their support in our health care services. Our clinical and research team includes autistic and gender diverse professionals and peer supporters. Thus, the relevance of our questions is ensured and this enables us to guarantee respect for all people involved.

The primary objective of our review and meta-analysis is to determine the prevalence of GD in individuals with a confirmed diagnosis of ASD according to the latest diagnostic criteria. In addition, our study aims to (1) identify factors that may influence the prevalence of GD within this population such as age, gender assigned at birth, type of ASD diagnosis, or type of GD and (2) enhance the understanding of GD within the context of autism, particularly the methods used to assess GD and the type of GD encountered within this population.

Methods

Protocol and registration

We followed the recommendations of the Preferred Reporting Items for Systematic Reviews and Meta-analyses[17] reporting guideline (see Supplementary Data S1). We registered the protocol of this review in PROSPERO (CRD42022320669).

Search strategy

On July 26, 2022, we conducted a comprehensive literature search across four main electronic databases (PubMed [including Medline], Web of Science [Clarivate], Embase [Elsevier], and Cochrane Library [Wiley]). We aimed to identify relevant literature published on or after January 2013, aligning with the DSM-5, which updated the diagnostic criteria and provided a more comprehensive and accurate framework for diagnosing and understanding ASD.

We used a combination of free-text and thesaurus terms for the relevant concepts. An information specialist (C.G.) helped us with the construction of the search algorithms (see Supplementary Data S2). We applied English and French language restrictions.

An automated alert for publication updates was set up to March 1, 2023, and we included all articles matching our inclusion criteria during this period.

We also checked all references of the selected articles; nevertheless, we did not identify and include additional studies in the review by this means.

Study selection

Two independent reviewers (G.B. and E.P.) conducted the literature search using Rayyan website (https://www.rayyan.ai/) to facilitate double-blind selection and to manage the duplicates. Each reviewer assessed the relevance of studies based on their titles and abstracts. Full texts were examined to determine eligibility. We consulted a third author (M.N.) to resolve any disagreements.

Inclusion criteria were as follows: (1) articles published after January 2013; (2) written in English or French; (3) focusing on individuals (or their parents) with a diagnosis of ASD according to DSM-5 criteria, established by a qualified health care professional and/or by a multidisciplinary team, and/or by specific assessment such as the ADOS-2 (Autism Diagnostic Observation Schedule—Second Edition[18]), the ADI-R (Autism Diagnostic Interview—Revised version[19]), or the DISCO (Diagnostic Interview for Social and Communication Disorders[20,21]); and (4) mentioning any kind of information about participants' gender. Eligible articles included original articles published in peer-reviewed journals, comprising prospective cohort studies, retrospective cohort studies, case–control studies, cross-sectional studies, qualitative studies, or clinical trials.

Exclusion criteria were as follows: (1) nonhuman population; (2) systematic reviews, narrative reviews, meta-analysis, case reports, editorials, posters, and conference abstract; (3) studies with a weak reliability of ASD diagnosis such as those involving self-diagnosis, the use of not-validated diagnosis scales, the use of screening tools, solely, or unspecified diagnosis method; (4) studies that only provided binary information on gender (male/female ratio).

Data extraction

For each included study, two authors (G.B.; E.P.) extracted the following variables using a standardized extraction form: (1) study characteristics: authors and year of publication, design of the study, prime objective of the study (linked with gender identity or not); (2) autistic population characteristics: use of a specific database for population recruitment or not, age distribution, number of ASD diagnosis, birth-assigned sex, type of ASD diagnosis (clinically validated diagnosis only, clinical diagnosis confirmed with specific psychometric assessments), presence of comorbidities (organic or psychiatric); and (3) GD characteristics: type of measure of gender identity (free self-declaration, parents declaration, multiple-choice questionnaire, specific psychometric item), number of gender diverse cases and their birth-assigned sex, type of GD, number of cases by type of GD if available.

Bias and quality assessment

The same two authors (G.B.; E.P.) assessed the methodological quality of the included studies using the Joanna Briggs Institute tool for prevalence studies (See Supplementary Data S3). This tool is designed to evaluate the methodological quality of studies and determine the extent to which a study has addressed potential biases in its design, conduct, and analysis. Each question in the tool was scored as "0" for "Yes" or "Not applicable" and "1" for "No" or "Unclear."

We calculated the total score for each article by summing its points. Based on this tool, we categorized studies as having low risk (scores 0–3), moderate risk (scores 4–6), or high risk (scores 7–9) of bias.

Statistical analysis

Author M.N. analyzed data using R software (version 4.1.2) with the "meta" package (version 6.5–0).

For estimation of prevalence of GD in the autistic population, we used random-effects models using the generalized linear mixed models with a logit transformation.

We assessed the heterogeneity between studies with the I^2 and the τ^2. We performed a planned meta-regression within age and a *post hoc* meta-regression within the year of publication. We performed *post hoc* subgroup analysis according to the following: (1) age; (2) year of publication; (3) gender assigned at birth; (4) type of measure of GD; (5) type of ASD diagnosis; (6) main objective of the study linked or not with gender; and (7) type of GD. We assessed publication biases by the asymmetry of the funnel plot. We did not perform multiple test correction in line with recommendations from the Cochrane Handbook.[22] Actually, adjustments for multiple testing are not routinely applied in systematic reviews because the primary goal is to summarize existing evidence rather than to test new hypotheses. Systematic reviews often involve multiple comparisons, but applying strict corrections like the Bonferroni method could lead to an overconservative approach, increasing the risk of type II errors (false negatives). Therefore, unless the review specifically aims to make new statistical inferences, corrections for multiplicity are not necessary. In our case, we aimed to aggregate existing evidence without introducing additional statistical constraints.

Results

Search result

We initially identified 7133 studies through the database search, with distribution as follows: PubMed (1792), EMBASE (2366), Web of Science (2860), and Cochrane (115). After removing duplicates and screening for relevance based on language and date criteria ($N = 3321$), we screened 3812 studies based on titles and abstracts, leading to the exclusion of 3376 irrelevant studies. From the remaining 436 full-text articles, we excluded 418 articles for not meeting the eligibility criteria outlined in the methods section. Therefore, the final sample comprised 24 studies, all in English, that met all eligibility criteria, with 18 identified through screening and 6 through scientific monitoring. Among these, 14 studies reported quantitative results on the prevalence of GD in the autistic population and were included in the meta-analysis (see flow chart in Fig. 1).

Systematic review

The overview of the data is provided in Table 1.

Among the 24 studies, 16 were primarily conducted with adults,[22–37] 5 with adolescents or young adults,[38–42] and 3 with children.[43–45] None of the studies focused on individuals with intellectual disability; however, a few articles mentioned the presence of psychiatric disorders, with anxiety disorders and depression being the most frequently cited.

Among the 24 articles selected, only 8 compared autism with the neurotypical population.[22,25,34,38,42–45] However, in all of these comparative studies, the proportion of individuals with GD was higher in the autistic population compared with the neurotypical population.

We observed wide heterogeneity among the studies, particularly concerning the definition of GD and its measurement. Various methods measured gender identity, including self-completed demographic questionnaires ($N = 16$),[23–25,27,28,31–34,36–38,40,42,43,46] self-completed specific item from questionnaires about gender, sexuality, and behavior ($N = 3$),[29,39,45] and parents' reports ($N = 3$),[26,30,35] and one article utilized both a self-completed specific questionnaire and a parents' report.[41] In addition, in one study, the question about gender identity emerged naturally during a semi-structured interview.[44]

In this review, we found that a wide range of terms and concepts are used to describe GD. Some articles were based on a binary vision of gender, in which the term "Transgender," "Transman," or "Transwoman" was used to indicate a perception of self-gender identity in the opposite gender of the one assigned at birth.[23,26–28,33,34,36,37,42,43,46] Other articles used a more fluid or non-binary concept of gender identity.[24,25,29,31,32,38,40,44,45] In those, people could define themselves in other terms, such as "Non-binary," "In between man and woman," "Agender," "Gender neutral," "Gender non-conforming," "Gender Queer," "Another gender identity," or "Questioning their gender identity." Finally, some studies considered participants as gender diverse when they answered "Sometimes," "Often," or "Very often" to the item: "I wish I was the opposite sex."[30,35,39]

Non-binary identities were the most reported identities in autistic people and therefore were mentioned in 18 different articles. Distinctive data between binary or non-binary gender identities were only available for 11 of these articles. Among these studies, 10 found non-binary genders to be more frequent than binary ones,[25,27,29,31–34,37,42,46] and the number of occurrences was equal in the last one.[23]

Meta-analysis

A total of 3894 autistic participants from 14 studies were included in the meta-analysis. Ten studies were excluded either because they did not provide quantitative data or because they used the same databases, thus potentially including the same subjects (in that case, we kept the study with the largest sample). Of the 14 studies, 8 were primarily conducted with adults, 4 with adolescents or young adults, and 2 with children. The analysis yielded a pooled prevalence estimate of GD in individuals with a formal diagnosis of ASD at 7.37%. The 95% confidence interval (CI) for the pooled prevalence estimate was [4.45; 11.98]. Figure 2 depicts the forest plot for this meta-analysis.

Results indicate a significant level of heterogeneity ($I^2 = 90\%$, Tau2 = 0.91, $p < 0.01$), with estimated prevalence ranging from 0.85% [0.02; 4.67] to 27.27% [18.32; 37.81].

Subgroup and meta-regression analyses. We hypothesized that age might influence the prevalence of GD in autism. However, the meta-regression analysis did not find any association between the age of the autistic population and

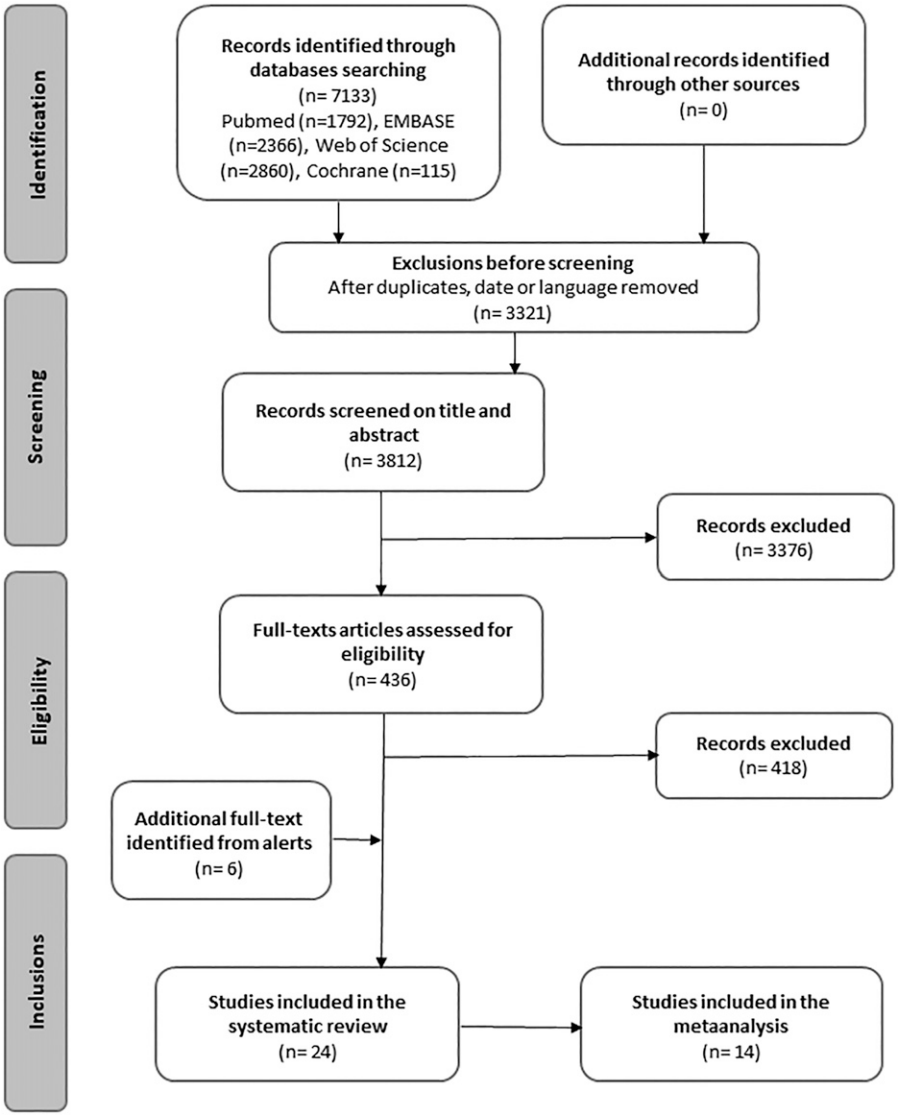

FIG. 1. Flow chart.

the prevalence of GD ($p = 0.72$). In addition, in a *post hoc* analysis, we evaluated the impact of the year of publication on GD prevalence, but found no significant association ($p = 0.83$).

We also expected the prevalence of GD in the autistic population to vary based on the gender assigned at birth. Our analysis revealed a higher prevalence of GD in individuals assigned female at birth (AFAB) (14.54%, 95% CI 6.85–28.24) compared with those assigned male at birth (AMAB) (8.15%, 95% CI 3.95–16.07). However, this difference was not statistically significant ($p = 0.18$) (see Supplementary Data S4, Supplementary Fig S4.1 in Supplementary Data S4).

The *post hoc* subgroup analysis did not reveal a significant difference in the prevalence of GD based on the type of measure used. The prevalence of GD was similar regardless of whether it was based on self-declaration (7.34%, 95% CI 4.40–12.01), a multi-choice questionnaire (10.99%, 95% CI 4.61–23.99), or a parent's declaration (2.79%, 95% CI 0.77–9.29) (see Supplementary Data S4, Supplementary Fig. S4.2 in Supplementary Data S4).

The *post hoc* analysis did not reveal a significant difference in the prevalence of GD based on the type of ASD diagnosis. Comparing studies where the diagnosis of ASD was clinical only (6.81%, 95% CI 3.73–12.11) with studies where the diagnosis was made using both a clinical assessment and psychometric measures (ADOS-2 and/or ADI and/or DISCO) (8.62%, 95% CI 3.51–19.66) did not yield a significant difference (see Supplementary Data S4, Supplementary Fig. S4.3 in Supplementary Data S4).

Contrarily, we observed a significant difference between studies where the main objective was linked to gender (13.71%, 95% CI 7.40–24.01) and studies where the main objective was not linked to gender (4.71%, 95% CI 2.76–7.92). The prevalence of GD was significantly higher in articles studying gender ($p < 0.01$) (see Fig. 3).

Finally, due to lack of specific data in most of the studies, we were unable to conduct a subgroup analysis distinguishing between binary or non-binary gender identities. Therefore, we could not quantify a significantly higher rate of non-binary identities versus binary identities as described in the systematic review (see Supplementary Data S4, Supplementary Fig. S4.4 in Supplementary Data S4).

Table 1. Overview of the Data

Study characteristics				*ASD population characteristics*				*Gender diversity characteristics*				
Authors (years)	*Ref number*	*Design of the study*	*Objectives of the study linked with gender*	*Database used for the population recruitment*	*Age of population—mean (SD) and [range]*	*Type of ASD diagnoses*	*Number of ASD cases (AFAB/AMAB)*	*Measure of gender diversity*	*Number of gender diversity cases (AFAB/AMAB)*	*Type of gender diversity*	*Inclusion in the meta-analysis*	*Risk of bias*
Bejerot & Eriksson (2014)	38	Case–control study	Yes	No	AMAB: 31.8 (7.8) [20–47]; AFAB: 28.1 (6.3) [20–47]	Established by a qualified health care professional AND ADOS-2	50 (24/26)	Self-completed demographic questionnaire	11 (8/3)	Answered "in-between man and woman" OR "transexual" OR "opposite sex" at the question "What is your gender identity?"	Yes	Low
Chang et al. (2022)	39	Cohort study	Yes	No	20.4 (2) [14–25]	Established by a qualified health care professional AND ADI-R	88 (9/79)	Item 29 of ASRI-4 (Adult Self-Report Inventory-4): "I wish I was the opposite sex."	24 (5/19)	Answered "sometimes" OR "often" OR "very often" at the question "I wish I was the opposite sex."	Yes	Low
Conner et al. (2022)	23	Cross-sectional study	No	No	14.87 (2.27) [12–20]	Established by a qualified health care professional AND ADOS-2	78 (16/62)	Self-completed demographic questionnaire	2 (1/1)	Non-Binary OR Transgender	Yes	Low
Cook et al. (2021)	24	Qualitative study	No	No	44.53 (12.03) [24–63]	Established by a qualified health care professional OR Established by a multidisciplinary team	17 (NA/NA)	Self-completed demographic questionnaire	3 (NA/NA)	Agender/Gender neutral	No	Low
Cook et al. (2022)	40	Qualitative study	No	No	44.53 (12.03) [25–64]	Established by a qualified health care professional OR Established by a multidisciplinary team	17 (NA/NA)	Self-completed demographic questionnaire	3 (NA/NA)	Agender/Gender neutral	No	Low
Cooper et al. (2023)	43	Cross-sectional study	No	No	17.6 (1.1) [15–22]	Established by a qualified health care professional	121 (36/82)	Self-completed demographic questionnaire	3 (NA/NA)	Another gender identity	Yes	Low
Corbett et al. (2023)	41	Case–control study	Yes	No	11.42 (1.03) [10–13]	Established by a qualified health care professional AND ADOS-2	140 (36/104)	Gender Diversity Screening Questionnaire self-report (GDSQ-S) AND Gender Diversity Screening Questionnaire parent-report (GDSQ-P)	NA	NA	No	Low
Folta et al. (2022)	25	Qualitative study	No	No	NA (NA) [18–23]	Established by a qualified health care professional	18 (NA/NA)	Self-completed demographic questionnaire	4 (NA/NA)	Agender/Non-Binary OR Transgender	No	Moderate
Graham Holmes et al. (2020)	26	Case–control study	No	Yes (IAN, Interactive Autism Network)	NA (NA) [18–30]	Established by a qualified health care professional	117 (22/95)	Parent report of their child's gender identity	1 (NA/NA)	Transgender/Non-binary	Yes	Low
Hedley et al. (2022)	27	Cohort study	No	No	41.75 (12.89) [20–71]	Established by a qualified health care professional	102 (59/35)	Self-completed demographic questionnaire	8 (NA/NA)	Non-Binary	Yes	Low
Huang et al. (2021)	28	Cross-sectional study	No	No	36.52 (13.92) [15–80]	Established by a qualified health care professional OR Established by a multidisciplinary team	657 (352/264)	Self-completed demographic questionnaire	37 (NA/NA)	Other	Yes	Low
Hull et al. (2021)	42	Cross-sectional study	No	Yes (CARD, Cambridge Autism Research Database)	NA (NA) [18–75]	Established by a qualified health care professional	305 (196/104)	Self-completed demographic questionnaire	23 (17/4)	Non-Binary OR Transgender	Yes	Low
Joyal et al. (2021)	29	Case–control study	Yes	No	19.2 (2.7) [NA-NA]	Established by a qualified health care professional	68 (27/41)	Question about gender identity in the Sexual Behavior Scale-Third edition (SBS-III)	24 (12/12)	Answered no at "I clearly identify myself as the gender that I was born (boy or girl)" OR Answered yes at "I consider myself as transgender"	Yes	Low

(*continued*)

TABLE 1. (CONTINUED)

Study characteristics				*ASD population characteristics*				*Gender diversity characteristics*				
Authors (years)	*Ref number*	*Design of the study*	*Objectives of the study linked with gender*	*Database used for the population recruitment*	*Age of population—mean (SD) and [range]*	*Type of ASD diagnoses*	*Number of ASD cases (AFAB/AMAB)*	*Measure of gender diversity*	*Number of gender diversity cases (AFAB/AMAB)*	*Type of gender diversity*	*Inclusion in the meta-analysis*	*Risk of bias*
Kanfiszer et al. (2017)	44	Qualitative study	No	No	NA (NA) [20–59]	ADOS-2 OR DISCO OR ADI-R	7 (7/0)	Semi-structured interviews in which emerged a "gender identity" category	4 (4/0)	Identified with a construct other than femininity, thus questioning their gender identity	No	Moderate
May et al. (2017)	30	Case–control study	Yes	Yes (NDAR, National Database for Autism Research)	NA (NA) [6–18]	ADOS-2	176 (33/136)	Item 110 of the CBCL (Child Behavior Checklist) "Wishes to be of opposite sex," completed by parent	7 (2/5)	Parent reports "sometimes" OR "often" at "Wishes to be of opposite sex"	Yes	Low
McQuaid et al. (2022)	31	Cross-sectional study	Yes	Yes (SPARK, Simons Foundation Powering Autism Research)	39.04 (13.59) [18–83]	Established by a qualified health care professional OR Established by a multidisciplinary team	651 (393/258)	Self-completed demographic questionnaire	67 (56/11)	Transfemale OR transmale OR gender nonconforming OR genderqueer OR another gender identity OR a combination of those responses	No	Low
McQuaid et al. (2023)	32	Case–control study	Yes	Yes (SPARK, Simons Foundation Powering Autism Research)	32.97 (8.7) [18–49]	Established by a qualified health care professional	502 (276/226)	Self-completed demographic questionnaire	62 (48/14)	Transfemale OR transmale OR gender nonconforming OR genderqueer OR another gender identity OR a combination of those responses	No	Low
Moseley et al. (2022)	33	Cross-sectional study	No	Yes (CARD, Cambridge Autism Research Database)	41.9 (13.4) [18–72]	Established by a qualified health care professional OR Established by a multidisciplinary team	314 (229/84)	Self-completed demographic questionnaire	55 (NA/NA)	Non-Binary OR Transgender	No	Low
Parenteau et al. (2023)	34	Qualitative study	No	Yes (SPARK, Simons Foundation Powering Autism Research)	26.51 (4.62) [NA-NA]	Established by a qualified health care professional	315 (150/165)	Self-completed demographic questionnaire	18 (NA/NA)	Non-Binary	No	Low
Pohl et al. (2014)	45	Case–control study	No	Yes (CARD, Cambridge Autism Research Database)	36.39 (11.98) [NA-NA]	Established by a qualified health care professional	320 (320/0)	Questions about gender identity in the Testosterone-related Medical Questionnaire (TMQ)	12 (12/0)	Answer "yes" at "Have you ever been diagnosed with gender identity disorder (GID)?" OR "Are you transsexual?"	Yes	Low
Strang et al. (2014)	35	Case–control study	Yes	No	12.21 (3.08) [6–18]	Established by a qualified health care professional AND ADOS-2 AND ADI-R	147 (24/123)	Item 110 of the CBCL (Child Behavior Checklist) "Wishes to be of opposite sex," completed by parent	8 (NA/NA)	Parent reports "sometimes" or "often" at "Wishes to be of opposite sex"	Yes	Low
Walsh et al. (2018)	36	Case–control study	Yes	No	44.67 (12.63) [16–80]	Established by a qualified health care professional	669 (347/322)	Self-completed "Gender Identity" multiple choice question: "man," "woman," "somewhat man, somewhat woman," "neither man nor woman," "I don't (yet) know," and "Other"	100 (75/25)	Non-Binary OR Transgender	Yes	Low

(*continued*)

TABLE 1. (CONTINUED)

Study characteristics				ASD population characteristics				Gender diversity characteristics				
Authors (years)	*Ref number*	*Design of the study*	*Objectives of the study linked with gender*	*Database used for the population recruitment*	*Age of population—mean (SD) and [range]*	*Type of ASD diagnoses*	*Number of ASD cases (AFAB/AMAB)*	*Measure of gender diversity*	*Number of gender diversity cases (AFAB/AMAB)*	*Type of gender diversity*	*Inclusion in the meta-analysis*	*Risk of bias*
Williams et al. (2023)	37	Cross-sectional study	No	Yes (SPARK, Simons Foundation Powering Autism Research)	37.41 (13.21) [18–79]	Established by a qualified health care professional	901 (569/332)	Self-completed demographic questionnaire	119 (92/27)	Non-Binary/Other OR Transgender	Yes	Low
Williams & Gotham (2021)	46	Cross-sectional study	No	Yes (SPARK, Simons Foundation Powering Autism Research)	30.89 (7.04) [18–45]	Established by a qualified health care professional	700 (441/259)	Self-completed demographic questionnaire	80 (61/19)	Non-binary/Other OR Transgender	No	Low

ADI-R, Autism Diagnostic Interview-Revised version; ADOS-2, Autism Diagnostic Observation Schedule-Second Edition; AFAB, assigned female at birth; AMAB, assigned male at birth; ASD, Autism Spectrum Disorder.

Bias management

Risk of bias in the systematic review. Using the Joanna Briggs Institute tool for prevalence studies, we identified two studies at moderate risk of bias[25,44] and 22 studies at low risk of bias (see Table 1). This indicates the overall good quality of the included publications and lends strength to our results.

Publication bias in the meta-analysis. To assess the impact of publication bias, we conducted a cumulative meta-analysis. The funnel plot, which checks for publication bias in meta-analyses, displayed an asymmetry of estimations, indicating potential publication bias (see Supplementary Data S5). This asymmetry suggested that studies reporting higher rates of GD in autistic people were underrepresented in the literature. To correct this potential bias, we performed a trim-and-fill procedure, which estimates the number of missing studies and adds them to the analysis to provide a more accurate estimate. In our case, we added four hypothetical studies. After this adjustment, the pooled prevalence of GD in the autistic population would have been 11.7% (95% CI 6.8%–19.2%).

Discussion

Prevalence of GD in autism

The main objective of our study was to estimate the prevalence of GD, taken in its broadest definition, in individuals with a clinically established diagnosis of ASD, and we found a prevalence of 7.37% [4.45–11.98].

It is in fact a really topical issue as shown by the recent increasing number of studies concerning gender in autism since 2020. Actually, we found 6 studies published between 2013 and 2019 included in our review and 18 since 2020. Among these 18 articles, 7 have been published in 2022 and 5 in 2023. Moreover, since this date, many articles recruit information about gender identity (other than a binary male/female sex ratio), even if the main objective of the study is not related to this specific question.

In international literature, the intersection between GD and neurodiversity has been mostly studied in people referred to GD clinics. For example, de Vries et al.[6] found an incidence of 7.8% of ASD diagnosis in a sample of children and adolescents referred to those clinics. In the adult population, the prevalence of clinically significant autistic traits has been measured at 5.5%, using screening questionnaires such as the AQ.[7] Finally, a recent meta-analysis showed that the prevalence of ASD diagnosis in the Gender Dysphoric/GID population was 11 times higher than the prevalence of autism estimate in the general population.[10]

A growing number of studies considering GD in autism suggest that the occurrence rate of GD is higher in autistic children[35] and adults[36] than in the neurotypical population. This meta-analysis covers an extensive selection of studies thus enabling us to measure a precise prevalence of GD in autism. These results contribute valuable insights to this important area of research. Moreover, it enables the inclusion of gender diverse individuals within its broad definition, moving beyond the restrictive frameworks imposed by diagnostic manuals such as the DSM and ICD. According to our results, the prevalence of GD in autistic people is 7.37% (4.45–11.98), which is significantly higher than the prevalence in the general population. Indeed, in his review, Zucker found

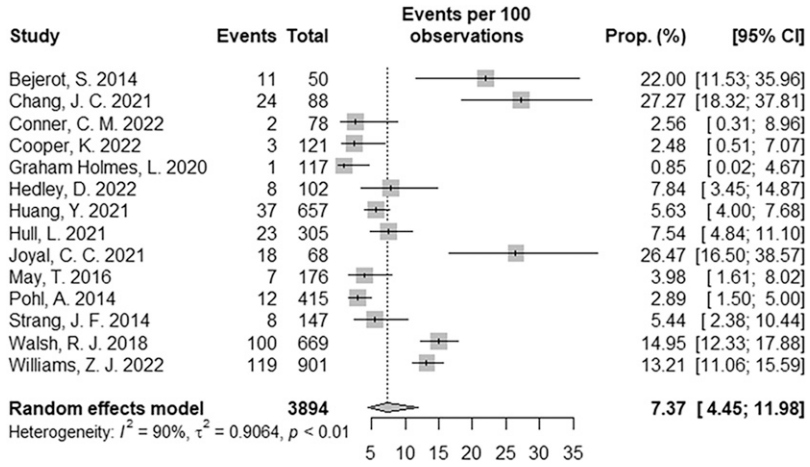

FIG. 2. Forest plot of the prevalence of GD in all studies included in the meta-analysis. GD, Gender Diversity.

that the prevalence of a self-reported transgender identity in children, adolescents, and adults ranged from 0.5% to 1.3%.[47] On his part, Zhang reviewed 19 articles in which the prevalence ranged from 0.5% to 4.5% among adults and 2.5% to 8.4% among children and adolescents, depending on the studies, when using a broader definition of GD.[48]

Factors influencing GD in autism

Another objective of our study was to identify factors that are concomitant or have an impact on the prevalence of GD in autism. Some studies have emphasized the influence of gender assigned at birth on GD prevalence among autistic individuals, the most cited study being Dewinter et al.[13] In this study assessing 675 autistic people, about 22% of AFAB and 8% of the AMAB reported "gender non-conforming feelings." In another recent study published in 2023, Khan et al. also indicated that youth with an ASD diagnosis and AFAB were more likely to have a diagnosis of gender dysphoria compared with youth with an ASD diagnosis and AMAB.[49] In addition, Jones et al. aligned with these results,[50] whereas other authors gainsaid this proposal.[2,6] In our meta-analysis, the difference in the prevalence of GD between individuals AFAB and AMAB within the autistic population did not reach statistical significance (14.54% [6.85–28.24] versus 8.15% [3.95–16.07]).

We searched for other factors that could affect the prevalence of GD in autism. Neither the age nor the type of ASD diagnosis or the type of measure of GD seems to impact this prevalence, according to subgroup analysis. In contrast, we found that GD prevalence was significantly higher when the main objective of the study was related to gender identity. Moreover, we found that gender identity is a pretty recent topic in scientific literature. In fact, the funnel plot found a significant publication bias in favor of an underestimation of the overall prevalence of GD. Thus, it is possible that gender diverse people are misidentified in some studies: for example, demographic questionnaires sometimes only focus on gender assigned at birth or do not allow to give any other answer than "man" or "woman," leading to an underestimation if not a marginalization of those who do not fit in these categories. Another explanation could be the exclusion of

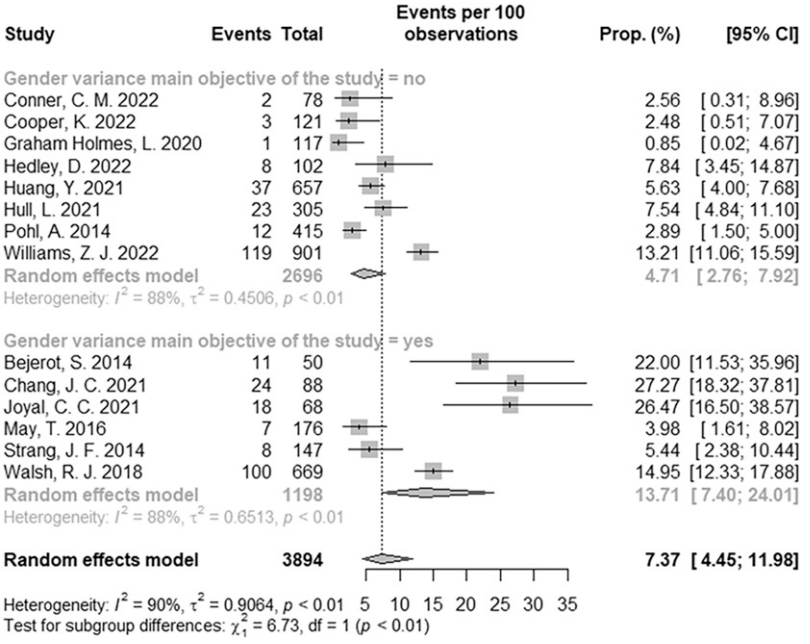

FIG. 3. Forest plot of the prevalence of GD according to main objective.

gender diverse individuals from study protocols. It is also possible that these individuals voluntarily opt out of taking part in research studies. However, it is challenging to substantiate these hypotheses. Moving forward, researchers should strive for greater inclusivity in representing the wide spectrum of gender identities among autistic people.

We also aimed to better define the types of GD encountered in autism. Beyond the high occurrence of Gender Dysphoria/GID, our study also highlights the broad spectrum of gender identities amongst autistic people. Although we could not calculate a statistical difference in the meta-analysis, our systematic review shows that the non-binary identities (meaning identities who were not encapsulated in "man," "transman," "woman," or "transwoman") seem to be particularly frequent, with a rich panel of terms and vocabulary such as the following: "nonbinary," "gender queer," "gender nonconforming," "in-between man and woman," "gender neutral," and "agender." As discussed earlier, Dewinter et al. found a rate of about 22% of AFAB and 8% of AMAB reporting "gender non-conforming feelings," whereas only 0.9% of the same group reported being transgender.[13] In this study, a single specific question recruited participants' gender identity. Participants were considered "gender non-conforming" when they answered the following: "partly male, partly female," "not male, nor female," "don't know (yet)," "different (e.g., human, no sex)," or "male" for people AFAB and "female" for people AMAB; participants were considered "transgender" only if they answered "male" for people AFAB and "female" for people AMAB.

This highlights the wide variety of words and concepts used in research exploring GD. Depending on the authors, it is difficult to determine whether "gender non-conforming feelings" truly reflect one's inner gender identity and thus indicate a situation of GD or if they instead depict feelings of incongruence with normative gender stereotypes. Another example in our review is the item "I wish I were of the opposite sex," used in some studies to explore gender identity. Responses such as "sometimes," "often," or "very often" are considered to be indicative of GD. However, these answers may rather reflect nonconformity with gender stereotypes or variability in gender expression.

Nevertheless, we did not find any study specifically examining the spectrum of gender identities in autism. Therefore, we needed more studies to determine whether non-binary identities are more common among autistic people and whether they identify themselves as transgender.

Theories on the link between GD and autism

To date, many different assumptions have been raised to explain greater variation in gender identity in autistic people. According to van der Miesen et al., all these theories could be divided into following three categories[2]: (1) the "biological" ones, which point to the influence of higher levels of antenatal testosterone exposure (known as "the Extreme Male Brain Theory") and to the assumption that GD and autism share common genetic patterns; (2) the psychological ones, which consider that psychological characteristics of autism, such as obsessions, rigidity, theory of mind impairments, or sensory processing, could influence the development of gender identity and explain GD in this population; and (3) the social ones, which propose that the effects of social experiences and notably the resistance to social norms, the stress of minorities, or the difficulties in social communication could explain GD in the autistic population. In a systematic review providing an overview of theories about the underlying mechanism of this association and the available evidence for and/or against them, Wattel et al. highlighted that all theories lack substantial empirical support.[51] Nevertheless, the most promising theories include the following: *the resistance to social norms*, which suggest that autistic individuals are less influenced by binary norm expectations compared with neurotypical individuals, and *the weakened sex differences*, which imply that the high prevalence of GD in autism may result from reduced differentiation between male and female traits among autistic individuals (e.g., autistic men exhibiting less masculine and more feminine traits and autistic women exhibiting less feminine and more masculine traits). The frequent report of non-binary identities by autistic people seems consistent with the social hypothesis, suggesting that non-binarity might arise from a divergence from traditional gender roles. Specifically, autistic people might experience their gender identity as more fluid and less constrained by conventional binary norms than neurotypicals due to their often unique engagement with social expectations. Although some studies seem to point in this direction, this remains a hypothesis more than a definitive conclusion. For instance, Walsh et al. found interest in the hypothesis of high resistance to social conditioning in autistic individuals regarding gender identity construction.[36] The higher GD in autism could be understood as a rejection of traditional gender-role norms, which combined with a below-typical concern for social norms could promote the disclosure of one's identity more freely.[36] The relationship between autism and non-binary identities is complex and influenced by various factors and requires further empirical investigation.

In our review, most of the studies did not mention any underlying hypothesis concerning the link between autism and GD. In fact, we found only five studies that quoted at least one of the hypotheses listed above. The assumption of the resistance to social norms has been cited thrice,[35,36,44] the extreme male brain theory has been cited twice,[35,38] and the prenatal androgen exposure has been only mentioned once.[45] Our study does not allow us to support either of these hypotheses, and more studies are needed to better understand the parallels between gender identity and the construction of self in the autistic population.

Limitations

This study is subject to several limitations that warrant consideration. First, the substantial heterogeneity observed among the included studies in terms of their design, sample characteristics, and measurement tools poses a challenge. Such diversity may introduce bias and restrict the generalizability of findings. Addressing this issue could involve advocating for standardized protocols and measurement tools across studies to enhance comparability.

Second, the variability in the definition and measurement of GD across studies presents a notable challenge. While some studies used binary frameworks, others embraced more fluid or non-binary concepts. This inconsistency complicates result comparisons and risks overlooking certain gender

identities within the autistic population. Efforts to establish clearer guidelines for defining and measuring GD could help mitigate this limitation.

Furthermore, the scarcity of comparative studies between autistic people and neurotypical populations limits our understanding of the differences in GD prevalence between these groups. Such comparative analyses are crucial for contextualizing findings and discerning unique patterns within the autistic population.

The presence of publication bias, indicated by the asymmetry of the funnel plot, is another notable limitation. This bias suggests that studies with higher rates of GD may be underrepresented in the literature, potentially skewing prevalence estimates and influencing interpretations of the findings. Implementing strategies such as the trim-and-fill procedure to correct for this bias may enhance the robustness of future meta-analyses. A search of gray literature could also help to better address this bias.

In addition, the predominantly cross-sectional nature of the included studies restricts our ability to infer causality or track changes in GD over time within the autistic population. Longitudinal studies would offer valuable insights into the trajectory of gender identity development in individuals with autism.

Finally, the exclusion criteria applied during the screening process may have introduced selection bias, as studies with a weak reliability of ASD diagnosis were omitted from the analysis. These studies may include individuals who do not have access to a comprehensive diagnostic process. These individuals may have distinct sociodemographic characteristics and, consequently, different experiences of gender identity compared with the population of our study.

Despite these limitations, the meta-analysis provides valuable insights into the prevalence of GD among autistic people. It underscores the need for further research to address the identified gaps and complexities in this field, ultimately advancing our understanding and support for autistic and gender diverse individuals.

Conclusions

It is important to consider the particularities of gender perception in autistic people to provide guidance to gender-affirming care practitioners who encounter them. In fact, the pathologization of autistic and transgender people could lead clinicians to see GD as a "symptom" of autism, which could prevent them from accessing proper gender-affirming care and delay their transitioning.[52]

The 8th edition of the Standard of Care from the World Professional Association for Transgender Health (SOC8 WPATH[53]) now suggests that health care professionals working with gender-diverse adolescents and children "receive training and develop expertise in autism spectrum disorders and other neurodevelopmental presentations or collaborate with a developmental disability expert." This recommendation aims at improving care for neurodivergent and gender-diverse adolescents and children. Given the delays and underdiagnosis of ASD, particularly in individuals without intellectual disabilities and especially those assigned female at birth, we believe this guidance is also pertinent for the adult population. By encouraging gender-affirming care professionals to recognize neurodiversity within the gender-diverse community, we can improve support for autism, helping them access tailored social skills training, mental health support, and peer support as needed.

In addition, it is possible that the non-binary gender identities often experienced by autistic people may confuse gender-affirming care practitioners more accustomed to binary transgender people. The SOC8 WPATH also provides recommendations regarding non-binary people and states that health care professionals should "provide nonbinary people with individualized assessment and treatment that affirms their experience of gender" and that they should "consider gender-affirming medical interventions (hormonal treatment or surgery) for nonbinary people in the absence of social gender transition" and "consider gender-affirming surgical interventions in the absence of hormonal treatment (...)." In the future, more studies focusing on GD in autism could help clinicians realize the interest and benefits of those recommendations when applied to the specific autistic population, thus improving the accessibility of gender-affirming care for autistic individuals.

Finally, we would like to emphasize the importance of pursuing extensive research on the intersection of autism and GD. It is crucial to ensure that the lived experiences of gender-diverse and neurodivergent individuals are not rendered invisible and that they are empowered to self-determine their gender identity.

Authorship Confirmation Statement

G.B. and E.P. conducted the literature search according to C.G.'s algorithm, and M.N. resolved disagreements and conducted statistical analysis. G.B. and E.P. drafted the initial version of the article. Subsequently, all authors (G.B., E.P., L.J., L.S., and C.D.) contributed to its completion, resulting in the current version. A.Z. conducted a final proofreading, paying particular attention to the terminology used. The article has been submitted solely to *Autism in Adulthood*.

Author Disclosure Statement

G.B., E.P., L.J., L.S., C.G., M.N., and C.D. have no interests to disclose. A.Z. is director of the *Fondation Agnodice* that aimed at supporting the integration and improving the well-being of transgender, non-binary, or gender-questioning individuals. She has no financial interest to disclose.

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Supplementary Material

Supplementary Data S1
Supplementary Data S2
Supplementary Data S3
Supplementary Data S4
Supplementary Data S5

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