

AUTISM AND GENDER DYSPHORIA: A NARRATIVE REVIEW OF THEIR COEXISTENCE AND CLINICAL IMPLICATIONS

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Abstract: Objective: Analyze the existing scientific literature in order to highlight and understand the coexistence of autism and gender dysphoria, as well as their clinical implications, in order to provide an updated and comprehensive overview of the topic.

Methods: Narrative bibliographic review through the PubMed database using the search terms “Autistic Disorder”, “Autism Spectrum Disorder” and “Gender Dysphoria”, in association with the Boolean operators “OR” and “AND”, initially resulting in 79 articles. After evaluation based on inclusion and exclusion criteria, only 16 articles became official sources for the present study.

Discussion: The studies reviewed highlight the importance of an integrated approach when examining the co-occurrence between Autism and Gender Dysphoria, in addition to exploring possible etiological connections with maternal exposure to testosterone during pregnancy. Furthermore, several social and behavioral factors, such as prejudice and cognitive inflexibility, influence the dynamics of belonging. The lack of family support and the lack of attendance at appointments emerge as some of the obstacles that hinder both the diagnosis and the promotion of mental health in affected individuals. **Final considerations:** Studies indicate that the coexistence between Autism and Gender Dysphoria is a significant challenge, requiring a personalized therapeutic approach. This complex interaction requires a deeper understanding to promote adequate diagnosis, treatment, mental health, well-being and emotional support.

Keywords: Autism Spectrum Disorder, Autism, Gender Dysphoria.

INTRODUCTION

Autism Spectrum Disorder (ASD) is a neurodivergence in which individuals face challenges in aspects of communication and socialization, in addition to manifesting

repetitive behaviors, as outlined by the American Psychiatric Association (2013). On the other hand, Gender Dysphoria (GD) is a disorder characterized by incongruence between the gender assigned at birth and the gender experienced by the individual, resulting in suffering, as well as difficulties in expression and socialization (DSM-5) (GLIDDEN D. et al., 2016). This condition is commonly experienced by the transgender or autistic population (GEORGE & STOKES, 2018; STRANG et al., 2021; VAN DER MIESEN AIR et al., 2018; COOPER K. et al., 2023). ASD and GD are relatively rare conditions, with prevalence rates estimated at 1 in 100 (BRUGHA et al. 2011) and 1 in 5,000 (Reed et al. 2009), respectively. The coexistence of ASD and GD implies additional complexity in understanding and expressing the identity of affected individuals, with significant implications for their psychosocial well-being (COLEMAN-SMITH RS et al., 2020). Furthermore, the young transgender population is susceptible to adverse psychiatric conditions, such as anxiety and depression (HISLE-GOMAN E. et al., 2019).

Studies have indicated a notable correlation between ASD and GD, where a disproportionately high incidence of autism or autistic traits has been observed in individuals with gender dysphoria. Specifically, 6.4% of children and 9.4% of adolescents referred for GD assessment (DE VRIES et al., 2010), as well as 5.5% of adults, manifested clinically significant symptoms of autistic traits (PASTERSKI et al, 2014). This correlation suggests the need for a more in-depth understanding and effective intervention strategies to improve the prognosis of these individuals.

The central objective of this review was to elucidate the complex interaction between Autism Spectrum Disorder (ASD) and Gender Dysphoria (GD), two disorders that

have profound implications for the quality of life, expression of identity and mental health of affected individuals. Through a critical and meticulous analysis of existing scientific literature, this study aimed to characterize the prevalence of ASD among individuals with GD and vice versa, providing a refined epidemiological understanding that can inform the medical and scientific community, as well as healthcare stakeholders. public. Finally, the study sought to identify and evaluate existing intervention strategies that aim to mitigate the adversities associated with the coexistence of ASD and GD, and propose evidence-based recommendations for a more effective and humanized clinical and therapeutic approach that can facilitate a more favorable prognosis. for affected individuals.

METHODOLOGY

This is a narrative bibliographic review based on the PVO strategy (Population, Variables, Outcome), used to develop the research through its guiding question: “How does the coexistence of autism and gender dysphoria manifest itself and what are its clinical implications? including challenges in diagnosis and treatment?”. In the context of this research, the population studied includes individuals diagnosed with autism and who also experience gender dysphoria. Its objective was to investigate the factors underlying the coexistence of these conditions, as well as to understand their clinical implications and the challenges related to diagnosis and treatment. The searches were carried out by searching the PubMed Central (PMC) database. The search terms were used in combination with the Boolean terms “OR” and “AND” through the following search strategy: ((Autistic Disorder) OR (Autism Spectrum Disorder)) AND (Gender Dysphoria). In total, 79 articles were found, which were subsequently evaluated based on selection criteria. The inclusion

criteria covered articles written in English, published between 2018 and 2023, related to the topics of research interest and which were reviews, meta-analyses, observational studies or case-control studies available in full. Duplicate articles, abstracts and studies that were not related to the research topic and did not meet the inclusion criteria were excluded. In the end, 16 articles were selected for analysis in the present study.

DISCUSSION

Autism spectrum disorder (ASD) is a neurodevelopmental condition characterized by significant challenges in social interaction and restricted and repetitive behavior patterns, essential criteria for its diagnosis. Gender diversity, which includes a wide range of identities that do not align with traditional gender expectations, encompasses individuals who explore or question their gender identities, including non-binary and transgender people. Gender dysphoria, which is the discomfort or suffering caused by an incongruity between the sex assigned at birth and the gender identity experienced, falls within this broader spectrum. The increase in ASD and GD co-occurrence rates is a current highlight, highlighting the potential impact on mental health in both conditions and making it essential to understand how this coexistence can influence diagnosis and treatment, in addition to to indicate the need for an integrated approach (HILTON G et al., 2022; KALLITSOUNAKI A.; WILLIAMS DM, 2022).

In recent years, there has been growing scientific interest in the intersection between ASD and GD. Systematic reviews, such as those carried out by Øien et al. (2018) and complemented by Nordahl-Hansen et al. (2019), have mapped a significant increase in publications and an evolution in the understanding of this coexistence since the

mid-20th century. A notable co-occurrence of ASD and gender dysphoria symptoms is observed across diverse cultures and age groups. Studies suggest that restricted and repetitive behaviors in childhood and adolescence are associated with the development of a diverse gender identity in adulthood (MAZZOLI F. et al., 2022). The presence of autistic traits, as reported by parents, appears to increase variation in gender identification (COLEMAN-SMITH RS et al., 2020).

Researchers such as Turban and van Schalkwyk (2018) propose that characteristics of ASD, such as cognitive inflexibility, may be related to the development of a divergent gender identity. This relationship is complex and multifaceted, with a significantly elevated prevalence of ASD in individuals with gender dysphoria, indicating an urgent need for in-depth understanding and adapted support approaches (COLEMAN-SMITH RS et al., 2020).

A retrospective case-control analysis involving thousands of children suggests that the diagnosis of GD is significantly more common among children with ASD compared to those without ASD (Hisle-Gorman et al., 2019). Hisle-Gorman et al., 2019 revealed that autistic children are 4.38 times more likely to experience GD compared to non-autistic children. The literature also explores how characteristics of ASD, such as difficulties with mentalization — understanding one's own and others' mental states — may relate to the experience of GD. Furthermore, rigidity in thinking, a trait associated with ASD, can influence how individuals interpret discrepancies between their behavior and the sex assigned at birth, which can lead to intense feelings of discomfort regarding one's gender identity (DE VRIES et al., 2010; JACOBS et al., 2014).

The study by Coleman-Smith RS et

al, (2020) identified a core category of “Conflict versus Congruence”, highlighting the multifaceted experience of conflict that participants experienced in relation to their body, interpersonal interactions, and psychological or intrapersonal conflict. It has been observed that as gender understanding and transition progress, individuals move from high conflict and low congruence to lower conflict and higher congruence. Autism was seen as a fundamental barrier to understanding and expressing authentic gender identities, exacerbating the impact of the social environment. However, for some, autism also acted as a protective factor, allowing them to live their gender identity authentically despite negative societal perceptions. The social environment generally represented a barrier, maintaining or increasing the feeling of conflict due to the lack of acceptance of neurodiversity and the dominance of cis-normativism. However, supportive environments, especially within the LGBT community, were essential in enabling exploration and understanding of gender and increasing personal congruence.

In the study by Cooper K. et al. (2023), participants described an interaction between the sensory needs of autism and negative experiences with the body, intensifying gender dysphoria. This was particularly pronounced during puberty. Furthermore, difficulties in managing changes also contributed to suffering during gender transition. There was significant convergence in views about how autism can worsen gender dysphoria and about clinical barriers to getting support. There were significant divergences between the groups when considering the role of autism in understanding one’s gender identity (COOPER K. et al., 2023).

Recognizing the complexity of this coexistence, Cooper et al. (2022) suggest carrying out longitudinal studies that follow

individuals from childhood to adulthood. This would allow for a detailed record of experiences of gender dysphoria in autistic people, offering both quantitative and qualitative insights.

Mazzoli F. et al. (2022) suggests a possible etiological link between ASD and GD, highlighting the theory of increased testosterone in amniotic fluid as a factor that may contribute to cognitive traits associated with autism. The correlation between ASD, Autistic Traits (AD) and GD with maternal exposure to testosterone during pregnancy is a topic of great relevance in medical research, with a significantly higher prevalence observed in males (HEYLENS G. et al., 2018). Furthermore, James W. and Grech V. (2020) propose three theoretical hypotheses to explain the relationships between autism, sex assigned at birth and exposure to testosterone. The first hypothesis proposes that one of the causes of autism is exposure to high levels of intrauterine testosterone. This hypothesis, initially formulated by Baron-Cohen, was confirmed by significant differences in amniotic fluid testosterone levels between cases and controls.

It is suggested that testosterone acts as a causal agent of autism, at least partially, before the 15th to 20th week of pregnancy. The second hypothesis proposed in the study suggests that high levels of maternal testosterone at the time of conception are positively associated with subsequent births of boys. This hypothesis was initially formulated by James and subsequently received additional support through reviews and meta-analyses. The theoretical basis of this hypothesis is that, in vertebrates (including humans), maternal testosterone levels influence fetal sex determination. According to this theory, higher levels of testosterone in the uterine environment at the time of conception increase the likelihood of conceiving a boy. The third

hypothesis addresses the greater frequency of gender dysphoria in cases of autism compared to controls. It is suggested that maternal stress, especially in the first trimester of pregnancy (when autism is believed to be programmed), may have a modest but significant effect on the sexual orientation of male offspring, and by inference, both autism and homosexuality. It may have been exposed to high levels of stress-induced maternal adrenal androgens, including testosterone

There is evidence that adults with ASD may experience an increased desire to be the opposite sex, related in part to the social isolation characteristic of ASD, which can influence the perception of belonging to their own gender (CHANG et al., 2022). Also indicating that adults with ASD who lacked family support during childhood and adolescence may present stereotypical behaviors and face mental health challenges, such as depression and anxiety, which significantly affect quality of life and may interact with GD experiences (CHANG et al, 2022). These factors are corroborated by studies that identified a higher incidence of bullying in autistic individuals who express a divergent gender identity (CRESSWELL et al., 2019).

Although there is robust literature on the coexistence of ASD and GD, diagnosis and therapeutic management remain challenging. Regarding the diagnosis, there are numerous complexities, such as the verbal expression necessary for the diagnosis of GD, which can be challenging for individuals with ASD, and the manifestation of atypical symptoms that can obscure the diagnosis and therapeutic approach (MIESEN et al., 2016). Diagnostic difficulties, including the possibility of false positives in previous studies, may contribute to the perception of a relationship between ASD and GD (HISLE-GOMAN E. et al., 2019). Furthermore, challenges in communication

and conception about gender in people with ASD can influence how symptoms are reported and interpreted, often based on information provided by parents and caregivers, which can introduce bias into the diagnostic process.

The “extremely masculine brain” theory proposes that people with ASD present a pattern of accentuated systematization and diminished empathy, which could explain some of the intersections observed between ASD and GD (MIESEN et al., 2016). And complex social factors also play a role in the development of GD in autistic individuals, including difficulties in social relationships and interpersonal understanding. Studies indicate that people with ASD can develop an atypical gender identity, which can increase the likelihood of GD experiences (TATENNO et al., 2008).

Social and economic aspects are also cited as influencing the observed coexistence of ASD and GD. For example, differences in access to diagnoses and treatments based on type of health insurance and ethnic origin have been observed, suggesting that factors such as income and insurance coverage may play a role. It was observed that co-occurring diagnoses of ASD and GD were more prevalent among young people whose sex recorded in the electronic medical record was female and among those who used private insurance. On the other hand, these diagnoses were less prevalent among young people of color, especially among black and Asian young people. Added to this, co-occurrence rates varied significantly based on sex recorded in the electronic medical record, race, and type of insurance, indicating potential disparities in ASD and GD diagnosis and service use. These results are important for developing and implementing strategies that aim to reduce these disparities and improve health outcomes for youth with co-occurring ASD and GD. Furthermore, young people diagnosed with

ASD who used private insurance were more likely to have a diagnosis of GD compared to those who used public insurance. This may be related to the correlation between income and insurance coverage, as well as variations in insurance coverage based on health needs and diagnoses. This situation is particularly relevant among youth with ASD who qualify for public insurance (such as Medicaid) but reside in states where gender-affirmative care is not explicitly covered by public insurance plans (KAHN NF et al., 2023).

Furthermore, changes in social standards regarding sexuality and gender identity may have contributed to an increase in GD diagnoses, particularly in the ASD population (ØIEN RA et al., 2018). The presence of the internet and social media may have fostered a more open discussion about gender issues, leading to a reduction in stigma and greater recognition and empowerment of diverse gender identities.

The coexistence of ASD and GD is of particular interest and clinical implication, as there may be an increased risk of mental health problems associated with affected patients, mainly depression and anxiety (KALLITSOUNAKI A.; WILLIAMS DM, 2022). The psychological stress associated with GD can lead to emotional difficulties and psychiatric disorders, especially among transgender individuals. These conditions can complicate treatment, highlighting the need for personalized and comprehensive therapeutic interventions. Furthermore, it is crucial to consider the possible presence of neurodivergences, as Autism Spectrum Disorder (ASD) can affect cognitive and emotional development (HILTON MN et al., 2022).

Autistic traits can contribute to the internalization of emotions and behaviors, increasing vulnerability to anxiety and depression in individuals with GD, negatively

affecting quality of life. According to Hilton, G et al. (2022), it is essential to identify and treat these traits to promote emotional well-being.

An emerging therapeutic approach is Gender Affirming Hormone Treatment (GAHT). A cross-sectional study using the Autism Spectrum Quotient (AQ) compared cisgender and transgender people before and after GAHT. The research found that, after one year of treatment, autistic transgender individuals showed a decrease in AQ scores, suggesting an improvement in well-being (MAZZOLI et al., 2022). Highlighting the importance of promoting studies that provide valuable insights into the interactions between autism, sex assigned at birth, prenatal exposure to testosterone, GD and other relevant factors. Understanding these dynamics can drive advances in research and treatment, contributing to reducing stigma and substantially improving the mental health of these individuals.

FINAL CONSIDERATIONS

The coexistence of Autism Spectrum Disorder (ASD) and Gender Dysphoria (GD) is a significant challenge in understanding the identity and promoting the mental health of affected individuals. A review of current studies reveals a notable correlation between ASD and GD, with a significant portion of people with GD showing autistic traits, and vice versa. This complex interaction highlights the need for a deeper understanding of these conditions and their interrelationships to improve the diagnosis, treatment and support offered to these individuals. The presence of autistic traits in people with GD can significantly influence their gender experience and therapeutic needs, requiring individualized support approaches. Furthermore, social, economic and cultural factors are fundamental in the manifestation and recognition of these

conditions, increasing the complexity of the interaction between ASD and GD.

The impact of these conditions on mental health is also a critical issue, with individuals often experiencing emotional challenges such as depression and anxiety. The effectiveness of Gender Affirming Hormone Treatment (GAHT) to improve the well-being of transgender individuals with autism is highlighted. However, despite the

vast literature on the coexistence of ASD and GD, studies focused on diagnosis and clinical management are still incipient and necessary, considering the complexities involved. An in-depth understanding of this interaction is vital to reducing stigma and improving mental health in these populations. Implementing personalized and comprehensive therapeutic interventions is essential for the well-being of affected individuals.

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