

Underdiagnosis in Female ASD: Limits of Diagnostic Instruments and the Urgency of Early Intervention

Subdiagnóstico no TEA feminino: limites dos instrumentos e a urgência da intervenção precoce

Subdiagnóstico en el TEA Femenino: Límites de los Instrumentos y la Urgencia de la Intervención Temprana

Isabella Ducarmo Leite¹, Pedro Melo de Queiroz², Altamiro Garcia Neto³, Gabriella Ficher de Assis Faria⁴, Lara Castro Caixeta⁵, Izabella Fernandes Menezes⁶, Daniel Alves Costa⁷, Talita Braga⁸

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1. Universidade Evangélica de Goiás (UniEvangélica). Anápolis, Goiás, Brasil. <https://orcid.org/0009-0005-6944-1538>

2. Universidade Federal de Goiás (UFG), Goiânia, Goiás, Brasil. <https://orcid.org/0009-0000-8616-5444>

3. Universidade Federal de Goiás (UFG), Goiânia, Goiás, Brasil. <https://orcid.org/0009-0007-8909-1058>

4. Pontifícia Universidade Católica de Goiás (PUC), Goiânia, Goiás, Brasil. <https://orcid.org/0009-0006-0431-0489>

5. Universidade Evangélica de Goiás (UniEvangélica). Anápolis, Goiás, Brasil. <https://orcid.org/0009-0001-5912-2235>

6. Universidade Evangélica de Goiás (UniEvangélica). Anápolis, Goiás, Brasil. <https://orcid.org/0009-0008-1499-3513>

7. Universidade Evangélica de Goiás (UniEvangélica). Anápolis, Goiás, Brasil. <https://orcid.org/0009-0003-3469-1479>

8. Universidade Evangélica de Goiás (UniEvangélica). Anápolis, Goiás, Brasil. <https://orcid.org/0000-0001-9794-7898>

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RESUMO

Objetivo: O Transtorno do Espectro Autista (TEA), envolve dificuldades de comunicação social e comportamentos repetitivos, com prevalência aproximada de 1% e histórico de maior incidência em homens. É observada a subdetecção feminina por exclusão das mulheres nos estudos. Diante disso, objetivou-se realizar revisão integrativa sobre desafios diagnósticos do TEA em mulheres e a importância da intervenção precoce. Trata-se de uma revisão integrativa realizada em seis etapas, com estratégia PICO e busca em julho/2025 na MEDLINE/PubMed com “autism”, “diagnosis” e “women”; inclusão: artigos em inglês/espanhol (2021–2025) sobre dificuldades diagnósticas, erros diagnósticos, diferenças entre gêneros e importância da intervenção; exclusão: revisões, causas/efeitos, tratamento exclusivo e fora do recorte. Após a aplicação dos critérios, 10 artigos foram analisados na íntegra. Instrumentos baseados em apresentações masculinas tendem a ignorar mulheres, que exibem interesses menos incomuns e maior atenção social, além disso há maior risco de traumas e outras condições de saúde mental, e a camuflagem contribui para diagnósticos perdidos. Logo, a identificação precoce pode mitigar riscos, ampliar acesso a serviços e favorecer identidade positiva visto que o reconhecimento tardio aumenta o sofrimento.

Palavras-chave: Autismo 1; Diagnóstico 2; Mulheres 3.

ABSTRACT

Objective: Autism Spectrum Disorder (ASD), involves difficulties in social communication and repetitive behaviors, with an approximate prevalence of 1% and a historically higher incidence in men. Female underdetection is observed due to the exclusion of women from studies. In view of this, the aim was to conduct an integrative review on diagnostic challenges of ASD in women and the importance of early intervention. This is an integrative review conducted in six stages, using the PICO strategy and a July/2025 search in MEDLINE/PubMed with “autism,” “diagnosis,” and “women”; inclusion: articles in English/Spanish (2021–2025) on diagnostic difficulties, misdiagnoses, gender differences, and the importance of intervention; exclusion: reviews, causes/effects, exclusive treatment, and out-of-scope studies. After applying the criteria, 10 articles were analyzed in full. Instruments based on male presentations tend to overlook women, who exhibit less atypical interests and greater social attentiveness; moreover, there is a higher risk of trauma and other mental health conditions, and camouflaging/masking contributes to missed diagnoses. Thus, early identification can mitigate risks, expand access to services, and foster a positive identity, whereas late recognition increases suffering.

Keywords: Autism 1; Diagnosis 2; Women 3.

RESUMEN

Objetivo: El Trastorno del Espectro Autista (TEA), implica dificultades de comunicación social y conductas repetitivas, con una prevalencia aproximada del 1% y una mayor incidencia histórica en hombres. Se observa subdetección femenina por la exclusión de mujeres en los estudios. Ante esto, se propuso realizar una revisión integrativa sobre los desafíos diagnósticos del TEA en mujeres y la importancia de la intervención temprana. Se trata de una revisión integrativa en seis etapas, con la estrategia PICO y una búsqueda en julio/2025 en MEDLINE/PubMed con “autism”, “diagnosis” y “women”; inclusión: artículos en inglés/español (2021–2025) sobre dificultades diagnósticas, errores diagnósticos, diferencias de género y relevancia de la intervención; exclusión: revisiones, causas/efectos, tratamiento exclusivo y estudios fuera del recorte. Tras aplicar los criterios, se analizaron íntegramente 10 artículos. Los instrumentos basados en presentaciones masculinas tienden a pasar por alto a las mujeres, que muestran intereses menos atípicos y mayor atención social; además, existe mayor riesgo de traumas y otros problemas de salud mental, y el camuflaje/masking contribuye a diagnósticos perdidos. Así, la identificación temprana puede mitigar riesgos, ampliar el acceso a servicios y favorecer una identidad positiva, mientras que el reconocimiento tardío incrementa el sufrimiento.

Palabras clave: Autismo 1; Diagnóstico 2; Mujeres 3.

Introduction

Autism Spectrum Disorder (ASD) is a neurodevelopmental condition characterized by early-onset social communication abnormalities and repetitive, stereotyped behaviors. The prevalence of ASD is approximately 1% worldwide. ASD was first described in 1943 by Leo Kanner, who believed it predominantly affected males. In 1944, Hans Asperger also described “childhood psychopathy,” later translated by Lorna Wing (1981) as Asperger Syndrome (1).

Physicians gradually identified the autistic group with more preserved intellectual and verbal abilities. However, this recognition highlighted the under-detection and diagnostic failures in girls with autism (2).

In this context, the incidence of ASD in males compared to females has been questioned in recent years. Since ASD screening tools are largely based on male presentations, many autistic females miss critical opportunities for early intervention and support (3)(4).

Compared to males, autistic females often face a higher number of traumas, an elevated risk for other mental health conditions, more disturbances in relationships, and challenges with self-identity (5). Additionally, females generally demonstrate higher levels of social attention, which may result in fewer “autistic symptoms” (6)(7). This becomes particularly concerning given ASD’s role as a psychopathological risk factor, leading to high levels of daily-life distress, including anxiety, depression, trauma, and suicidal thoughts, especially in milder forms of ASD (8).

Given these diverse issues, this integrative review was conducted, focusing on diagnostic challenges and the importance of early intervention.

Method

This is an integrative literature review (9). Initially, the guiding question was defined using the PICO strategy. In this research, the “population” was women on the autism spectrum. The “interest” was the diagnostic challenges and the importance of early intervention. The “context” included the different clinical manifestations across genders. The research question was: *What are the diagnostic difficulties and therapeutic importance of ASD in women beyond stereotypical manifestations of the disorder?*

The searches were conducted in July 2025 in the National Library of Medicine National Center for Biotechnology Information (MEDLINE/PubMed). Health science descriptors (DeCS/MeSH) used were: “autism” AND “diagnosis” AND “women.”

Inclusion criteria were: articles in English or Spanish, published between 2021 and 2025, addressing diagnostic challenges in autistic women, common diagnostic errors, conditions in which women were diagnosed prior to ASD, gender differences in clinical presentation, and the importance of early intervention. Exclusion criteria included: reviews, studies on causes and effects of autism, treatment-focused studies, and articles

outside the temporal scope. The PRISMA strategy (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) (10) was applied in the selection and exclusion stages. From 1,019 results, 10 articles were fully analyzed after applying the inclusion and exclusion criteria.

Results and Discussion

ASD is a neurodevelopmental syndrome characterized by difficulties in social reciprocity, social communication, flexibility, and sensory processing. Early identification can mitigate some of these risks and improve quality of life. However, the lack of recognition of autism in females contributes to their mental suffering (11)(12), as for many years, ASD screening and diagnostic tools were largely based on male presentations, causing females to be overlooked or mischaracterized. The term “Asperger Syndrome” was also erroneously used to describe females with more preserved intellectual and verbal abilities, even after its removal from the DSM-5 in 2013 (8)(13). This led to significant losses in access to interventions and early support. Only upon receiving a diagnosis could these women make sense of their struggles, reframing their lives through a diagnostic “lens” (14)(15).

Different Clinical Manifestations

It is important to note that girls and boys face different social criteria, and girls may express their autistic symptoms differently from male patterns. The Female Phenotype Theory (FPT) suggests that autistic females often present less obvious social impairments than autistic males, with lower severity of externalizing behaviors and higher internalization of emotional difficulties (13).

Moreover, autistic females tend to demonstrate greater empathy and social functioning, exhibit fewer restricted and repetitive behaviors and interests (RRBIs), higher social motivation, better communication skills, and a lower number of repetitive behaviors. They often have more friends (1)(2)(3).

Age of “Red Flags” and Pre-Diagnosis

Another important factor is the age of diagnosis. Although ASD is more easily identified in childhood, diagnosis is often later in adult life for females. Autistic females receive an ASD diagnosis at an average age of 23.57 years, compared to 16.92 years for autistic males. This difference was significant. In another study, the mean age of autism diagnosis was higher in females (14.6 years) than in males (12.2 years) (16)(13). Females diagnosed later report a higher number of traumas, greater risk of mental health problems, increased difficulty maintaining employment, and higher rates of sexual abuse, bullying, and mental harassment. They expressed sadness considering how different their lives could have been with timely ASD diagnosis (5)(7)(14).

Female struggles are also exacerbated by hormonal changes, with perimenopause frequently triggering a search for a diagnosis after decades of unexplained struggles and trauma. Perimenopause can worsen symptoms and increase anxiety and sadness, causing “brain fog” (17).

Camouflaging or Masking

Over time, many females develop the ability to “mimic” neurotypical patterns through social insights, such as suppressing stereotyped behaviors, forcing eye contact, and using rehearsed verbal and non-verbal repertoires to fit into society. This is known as camouflaging or masking (3)(4).

Females may appear to be “functioning well” until, at a certain age, social demands – such as academic performance, workplace context, and interaction challenges – exceed their masking capacity. The struggle for accessibility, before and after an autism diagnosis, is exhausting, as they strive to hide difficulties or explain why they do not fit typical expectations (14).

Diagnosis of Other Mental Disorders Before ASD

A growing number of studies highlight the likelihood of patients with ASD being erroneously diagnosed with other conditions due to phenotypic overlap. 54.2% of autistic females and 40.9% of autistic males received at least one psychiatric diagnosis prior to an autism diagnosis, with ADHD, anxiety, and depressive disorders being most common (16). The most frequent misdiagnosis for females was personality disorder (36.4%), especially Borderline Personality Disorder (BPD). Other conditions commonly confused with ASD or occurring as comorbidities include anxiety, depression, Obsessive-Compulsive Disorder (OCD), and Attention Deficit/Hyperactivity Disorder (ADHD) (2)(13).

Final Considerations

Evidence demonstrates that females with ASD often present symptoms distinct from males and frequently mask their autistic behaviors to conform to social expectations. This camouflaging results in delayed or incorrect diagnoses, negatively impacting quality of life. Male predominance in autism and expectations of male behavior patterns contribute to the underrepresentation of females in diagnoses. Additionally, the tendency to diagnose autistic females with other mental disorders before considering ASD highlights the need for a more gender-sensitive diagnostic approach.

Therefore, it is essential to develop screening tools adapted to female ASD characteristics and train healthcare professionals to recognize these nuances. Early and appropriate interventions can significantly improve the quality of life of autistic females. It is also important to note that both genders share some diagnostic parameters, and individual differences sometimes outweigh gender-specific patterns.

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I am grateful to my family, psychiatrist, and psychologist for enabling my access to diagnosis and treatment, an opportunity I recognize is not available to everyone. The time between the first consultation and final diagnosis marked my journey and highlights how delays in diagnosis remain a concerning reality for many females. This experience supports and motivates this work, which aims, with rigor and sensitivity, to contribute to earlier recognition and appropriate care.

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Correspondent author:

Isabella Ducarmo Leite

Alameda E-3 quadra 5 lote 28 jardins Mônaco, bairro

Vera Cruz, CEP: 74934-682.

Aparecida de Goiânia, Goiás, Brasil.