




DIAGNOSTIC CRITERIA FOR AUTISM SPECTRUM DISORDER (ASD): COMMUNICATION DEFICITS AND RESTRICTIVE PATTERNS

CRITÉRIOS DIAGNÓSTICOS PARA O TRANSTORNO DO ESPECTRO AUTISTA (TEA): DÉFICITS DE COMUNICAÇÃO E PADRÕES RESTRITIVOS

CRITERIOS DIAGNÓSTICOS PARA EL TRASTORNO DEL ESPECTRO AUTISTA (TEA): DÉFICITS DE COMUNICACIÓN Y PATRONES RESTRICTIVOS

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ABSTRACT

Autism Spectrum Disorder (ASD) is a complex and multifactorial neurodevelopmental condition characterized by persistent deficits in communication and social interaction, as well as restricted and repetitive patterns of behavior, interests, or activities. This study consisted of a narrative literature review aimed at synthesizing contemporary scientific evidence on the diagnostic criteria for ASD, with a focus on communication deficits and restrictive patterns, using the PubMed database. Current criteria (DSM-5 and ICD-11) converge into two main domains, highlighting, in communication, the lack of socioemotional reciprocity and impairment in joint attention. In the behavioral domain, motor stereotypies, insistence on inflexible routines, and sensory hyper- or hyporeactivity are notable. Diagnosis, which remains essentially clinical despite advances in neurobiological and genetic research, relies on gold-standard tools such as ADOS-2 and ADI-R. It is concluded that early detection and individualized, multidisciplinary management, including interventions such as ABA, ESDM, JASPER, and the use of Augmentative and Alternative Communication (AAC), are crucial to promote neuroplasticity, improve functional prognosis, and enhance the quality of life of individuals with ASD.

Keywords: Autism Spectrum Disorder. Diagnostic Criteria. Social Communication. Restrictive Patterns. Early Intervention.

RESUMO

O Transtorno do Espectro Autista (TEA) é uma desordem do neurodesenvolvimento complexa e multifatorial, caracterizada por déficits persistentes na comunicação e interação social, além de padrões restritos e repetitivos de comportamento, interesses

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ou atividades. Este estudo constituiu-se como uma revisão bibliográfica narrativa com o objetivo de sintetizar as evidências científicas contemporâneas sobre os critérios diagnósticos do TEA, com foco nos déficits de comunicação e padrões restritivos, utilizando a base de dados PubMed. Os critérios atuais (DSM-5 e CID-11) convergem em dois domínios principais, destacando-se, na comunicação, a falta de reciprocidade socioemocional e o prejuízo na atenção compartilhada (joint attention). No domínio comportamental, são notáveis as estereotipias motoras, a insistência em rotinas inflexíveis e a hiper/hiporreatividade sensorial. O diagnóstico, que permanece essencialmente clínico apesar dos avanços neurobiológicos e genéticos, utiliza ferramentas padrão-ouro como o ADOS-2 e ADI-R. Conclui-se que a detecção precoce e o manejo individualizado e multidisciplinar, incluindo intervenções como ABA, ESDM, JASPER e o uso de Comunicação Aumentativa e Alternativa (CAA), são cruciais para favorecer a neuroplasticidade, melhorar o prognóstico funcional e a qualidade de vida dos indivíduos com TEA.

Palavras-chave: Transtorno do Espectro Autista. Critérios Diagnósticos. Comunicação Social. Padrões Restritivos. Intervenção Precoce.

RESUMEN

El Trastorno del Espectro Autista (TEA) es un trastorno del neurodesarrollo complejo y multifactorial, caracterizado por déficits persistentes en la comunicación e interacción social, así como por patrones restringidos y repetitivos de comportamiento, intereses o actividades. Este estudio consistió en una revisión bibliográfica narrativa con el objetivo de sintetizar la evidencia científica contemporánea sobre los criterios diagnósticos del TEA, con énfasis en los déficits de comunicación y los patrones restrictivos, utilizando la base de datos PubMed. Los criterios actuales (DSM-5 y CIE-11) convergen en dos dominios principales, destacándose, en la comunicación, la falta de reciprocidad socioemocional y el deterioro en la atención conjunta (joint attention). En el dominio conductual, son notables las estereotipias motoras, la insistencia en rutinas inflexibles y la hiper/hiporreatividad sensorial. El diagnóstico, que sigue siendo esencialmente clínico a pesar de los avances neurobiológicos y genéticos, utiliza herramientas de referencia como ADOS-2 y ADI-R. Se concluye que la detección temprana y el manejo individualizado y multidisciplinario, incluyendo intervenciones como ABA, ESDM, JASPER y el uso de la Comunicación Aumentativa y Alternativa (CAA), son fundamentales para favorecer la neuroplasticidad, mejorar el pronóstico funcional y la calidad de vida de las personas con TEA.

Palabras clave: Trastorno del Espectro Autista. Criterios Diagnósticos. Comunicación Social. Patrones Restrictivos. Intervención Temprana.



1 INTRODUCTION

Autism Spectrum Disorder (ASD) is a heterogeneous neurodevelopmental disorder characterized by persistent impairments in social communication and social interaction, as well as the presence of restricted and repetitive patterns of behavior, interests, or activities (Genovese & Butler, 2023). It is a highly heterogeneous condition, both in its clinical presentation and in its etiology, affecting approximately 2% of the child population and with a higher prevalence in males (Genovese & Butler, 2023). The transition to the concept of "spectrum" reflected the diversity of clinical presentations, ranging from individuals with high abilities and functional language to cases with severe cognitive impairment and absence of speech (Lin et al., 2022; Lamanna & Meldolesi, 2024).

The etiology of ASD is multifactorial, involving a complex genetic architecture and environmental interactions that modulate brain connectivity during critical developmental windows (Genovese & Butler, 2023). Clinically, early diagnosis is critical for the implementation of interventions that promote neuroplasticity and functionality. However, the identification of early signs is often hampered by overlap with other conditions, such as Attention Deficit Hyperactivity Disorder (ADHD), and the need for high-sensitivity diagnostic tools that assess the fundamental domains of communication and behavior (Velarde & Cárdenas, 2022; Anixt et al., 2024).

2 METHODOLOGY

The present study is a bibliographic review of a narrative nature, structured with the objective of synthesizing contemporary scientific evidence on the diagnostic criteria of ASD, focusing on communication deficits and restrictive patterns. Data mining was performed in the PubMed database, using the descriptors "Autism Spectrum Disorder", "Child" and "Diagnosis", integrated according to the Medical Subject Headings (MeSH) terminology. The selection covered articles published in the last five years (2020-2025), available in full and written in English or Portuguese. Exclusion criteria were applied to remove studies focused exclusively on animal models with no human clinical correlation, duplicate publications, and reviews of low methodological rigor. The curatorial process involved the screening of titles and abstracts, followed by the critical reading of the full texts to validate their diagnostic relevance. The information extracted was organized in a descriptive way.



3 RESULTS

The current diagnostic criteria, based on the DSM-5 and ICD-11, converge on two main domains. In the domain of communication and social interaction, deficits are manifested by a lack of socio-emotional reciprocity, difficulties in initiating or maintaining conversations, and limited use of non-verbal behaviors, such as eye contact and gestures (Lin et al., 2022; Anixt et al., 2024). The absence or impairment of shared attention is a critical early marker for diagnosis in young children, being predictive of language severity in the future (Anixt et al., 2024).

In the domain of restrictive and repetitive patterns, the results indicate a range of behaviors that includes motor stereotypies, insistence on inflexible routines, highly fixed interests and abnormal in intensity or focus, as well as hyper- or hyporeactivity to sensory stimuli (Genovese & Butler, 2023; Lamanna & Meldolesi, 2024). This sensitivity to specific sounds or textures is a relevant clinical criterion, where sensory reaction can trigger challenging behaviors and *meltdowns*, directly impacting daily functioning and participation in educational activities (Anixt et al., 2024). Neuroimaging and neurobiology studies suggest that these behaviors are associated with changes in white matter integrity and functional connectivity of areas such as the prefrontal cortex and superior temporal sulcus (Lamanna & Meldolesi, 2024).

The gold standard diagnostic tools include the ADOS-2 (*Autism Diagnostic Observation Schedule*) and the ADI-R (*Autism Diagnostic Interview-Revised*), which allow for a structured assessment of symptoms (Lin et al., 2022). In initial screening settings in Primary Care, M-CHAT-R/F is widely recommended for children between 16 and 30 months (Anixt et al., 2024). In addition, the literature highlights that in cases of comorbidity with ADHD, the diagnosis of ASD can be postponed by up to two years, since inattention and impulsivity can mask primary social deficits (Velarde & Cárdenas, 2022).

Neurobiological and genetic evidence corroborates these diagnostic criteria, revealing an estimated inheritance of between 70% and 90% (Genovese & Butler, 2023). Changes in dendritic spine architecture and synaptic connectivity, particularly in the cortex, are associated with the observed deficits in social cognition and executive control (Lamanna & Meldolesi, 2024). Chromosomal microarray studies have identified that deletions or duplications in regions such as 15q11.2 and 16p11.2 are the most common cytogenetic anomalies in individuals with ASD, suggesting that these genetic variations



directly influence neuronal signaling pathways that underpin social behavior and cognitive flexibility (Genovese & Butler, 2023).

For children with significant verbal limitations, the use of Augmentative and Alternative Communication (AAC) systems is essential, with the *Picture Exchange Communication System* (PECS) and speech-generating devices (SGDs) proving superior to the use of sign language in developing fundamental skills such as the act of prompting. (Anixt et al., 2024)

Studies indicate that PECS significantly improves the ability to interact socially, being a priority recommendation for individuals who have failures in other speech approaches (Lin et al., 2022). The effectiveness of these tools is maximized when interventions are integrated into specific and contextual daily routines, such as mealtime, facilitating functional learning (Anixt et al., 2024).

4 DISCUSSION

The discussion of ASD diagnosis emphasizes that despite advances in the identification of biomarkers and genetic variants (such as mutations in cell adhesion genes), diagnosis remains essentially clinical (Genovese & Butler, 2023). The heterogeneity of the spectrum requires the clinician not only to identify the presence of symptoms, but to grade the level of support required (Level 1, 2, or 3), which directly impacts therapeutic planning (Anixt et al., 2024).

The integration of arts and music-based interventions has shown potential to mitigate social communication deficits by strengthening auditory-motor connectivity and facilitating the expression of emotions that the verbal channel often does not reach (Bernier et al., 2021). However, the "diagnostic vacuum" in minority populations or those with subtle presentations (female phenotype) remains a challenge, requiring clinicians to be on the lookout for "social camouflage" strategies that may conceal restrictive patterns (Lin et al., 2022).

It is concluded that the assertive diagnosis of ASD depends on a multidisciplinary look that considers the child's developmental trajectory. Timely detection of failures in social reciprocity and identification of repetitive behaviors allow the child to access evidence-based therapies (such as ABA, ESDM, or JASPER) early, significantly improving functional prognosis and family quality of life (Anixt et al., 2024; Lin et al., 2022).



5 CONCLUSION

Autism Spectrum Disorder is a complex and multifactorial neurobiological condition, resulting from the interaction between genetic, epigenetic, and environmental factors, which directly impact the development and functionality of neural circuits (Genovese & Butler, 2023; Lamanna & Meldolesi, 2024). The broad clinical heterogeneity observed reflects both the diversity of mechanisms involved and the different forms of phenotypic expression of the disorder, often associated with psychiatric and behavioral comorbidities that amplify its diagnostic and therapeutic complexity (Genovese & Butler, 2023).

Recent advances in the understanding of the neurobiological mechanisms of ASD, especially with regard to synaptic changes and gene regulation, have contributed to a more integrated view of the disease, although there are still important gaps in the understanding of its pathophysiology (Lamanna & Meldolesi, 2024). In this context, the relevance of early diagnosis is highlighted, which enables the implementation of interventions in critical phases of brain development, favoring better functional outcomes (Anixt et al., 2024).

The management of ASD must necessarily be individualized and multidisciplinary, based on interventions with scientific evidence, including behavioral approaches, language therapies, and strategies aimed at the development of adaptive skills (Anixt et al., 2024). Complementary interventions, such as artistic activities, also show promise by favoring communication, social interaction, and emotional regulation, expanding therapeutic possibilities (BERNIER et al., 2021).

Thus, the approach to ASD requires not only the recognition of its complexity, but also the integration between scientific knowledge, clinical practice, and family support, with the aim of promoting the global development, autonomy, and quality of life of affected individuals.

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