

International Journal of Health Science

Acceptance date: 05/03/2025

Submission date: 25/02/2025

THE ROLE OF EARLY DIAGNOSIS IN AUTISM: IMPACTS ON QUALITY OF LIFE AND CHILD DEVELOPMENT

Carina Toledo Scoparo Barioni

Positivo University. Faculty of Medicine
Curitiba-Paraná

<http://lattes.cnpq.br/8603504137422287>

Nilene Sales

Positivo University. Faculty of Medicine
Curitiba - Paraná

<https://orcid.org/0009-0009-2788-9800>

Renata Paes de Barros Wandresen

Positivo University. Faculty of Medicine
Curitiba - Paraná

<http://lattes.cnpq.br/9442494633948603>

Bárbara Bruna de Araújo Oliveira Kubo

Positivo University. Faculty of Medicine
Curitiba - Paraná

<https://lattes.cnpq.br/4095152668763776>

Amanda Franceschi Coimbra

Positivo University. Faculty of Medicine
Curitiba - Paraná

<https://orcid.org/0009-0007-8579-3300>

Luiza Myuki Saita

Positivo University. Faculty of Medicine
Curitiba - Paraná

<https://orcid.org/0009-0008-0478-526X>

João Victor Campos Ávila da Silva

Positivo University. Faculty of Medicine
Curitiba - Paraná

<https://lattes.cnpq.br/3621281997549378>

All content in this magazine is
licensed under a Creative Com-
mons Attribution License. Attri-
bution-Non-Commercial-Non-
Derivatives 4.0 International (CC
BY-NC-ND 4.0).



Lucas Pasquini Severo

Positivo University. Faculty of Medicine
Curitiba - Paraná
<https://orcid.org/0009-0000-1435-0886/>

Luana Vello

Positivo University. Faculty of Medicine
Curitiba - Paraná
<https://orcid.org/0009-0003-2559-6250>

Rodolfo Gavilak Drozino

Positivo University. Faculty of Medicine
Curitiba - Paraná
<https://orcid.org/0009-0004-1036-6820>

Gustavo Pichel Kakuda

Positivo University. Faculty of Medicine
Curitiba - Paraná
<https://orcid.org/0009-0004-8741-0420>

Altair Rogério Ambrosio

Positivo University. Department of Medicine
Curitiba-Paraná
<http://lattes.cnpq.br/4059028344359510>

Abstract: Introduction: Early diagnosis of Autism Spectrum Disorder (ASD) has been recognized as essential for improving quality of life and child development, allowing for specific interventions to improve social, communication and behavioral skills. Due to the high complexity and variability of symptoms, diagnostic strategies and access to specialized interventions have become the focus of recent research. **Objectives:** This article aims to analyze the effectiveness of early interventions in the development of children with ASD, seeking to highlight the benefits in independence and social integration provided by early diagnosis and treatment. **Methodology:** This is a systematic review, conducted in the Medline and PubMed databases, selecting articles between 2019 and 2024, using terms such as “autism”, “early diagnosis” and “quality of life”. **Results:** It was observed that the use of tools such as M-CHAT facilitates early screening, but access to diagnosis still faces socioeconomic and cultural barriers. The review suggests that early diagnosis, combined with inclusive public policies, expands children’s development possibilities and improves family support. **Conclusion:** It is concluded that early diagnosis and individualized interventions in ASD are fundamental for children’s social and behavioral progress and that equitable public policies are essential to ensure accessibility and continuity of care. **Keywords:** Autism spectrum disorder (ASD); autism; child; early diagnosis; treatment; quality of life;

INTRODUCTION

Autism Spectrum Disorder (ASD) is a complex neurobiological condition that affects the social, communication and behavioral development of individuals from early childhood. The prevalence of autism has increased significantly in recent decades, making it a central public health concern. Early detection of ASD is essential in order to initiate interventions

that can mitigate the challenges associated with the disorder and improve the developmental trajectory of diagnosed children [1].

Diagnosing ASD is a challenging process, given the broad spectrum of manifestations and the variability in the intensity of symptoms. Generally, the diagnosis is clinical, based on observations of the child's behavior and reports from caregivers, with signs such as communication difficulties, repetitive behaviors and limited social interaction usually appearing before the age of three. Recently, advances in screening tools, such as the M-CHAT (Modified Checklist for Autism in Toddlers), have facilitated early detection, allowing for more effective actions [2].

In this scenario, early interventions, carried out in the first years of life, have been shown to be effective in promoting social, cognitive and communication skills in children with ASD. These interventions include behavioral, speech and occupational therapies, which are customized to meet the child's individual needs. Current literature highlights the importance of accurate and early diagnosis, as the earlier ASD is identified, the sooner children can be referred for interventions that improve their developmental prospects [3].

The quality of life of children with ASD and their families is also directly influenced by how early and appropriate the interventions they receive are. Early interventions not only help in the acquisition of functional skills, but also minimize family stress and promote more effective social integration. Consequently, the implementation of early diagnosis and intervention programs is a priority in health policies to improve the quality of life of these children and their families [4].

The main objective of this study is to analyze the effectiveness of early interventions in the social, communicational and behavioral development of children with autism spectrum disorder. In addition, it seeks to explore the benefits of these interventions, especially

in promoting greater independence and social integration throughout these children's lives. The study also aims to provide scientific evidence that can guide both clinical practices and the development of public policies aimed at the care and inclusion of children with ASD, offering subsidies to improve family and social support, highlighting the importance of an integrated and continuous approach from the first years of life

METHODOLOGY

This article is a systematic review conducted following the PRISMA strategies (P: Preferred, R: Reporting, I: Items, S: Systematic, R: Reviews, M: Meta-Analyses) to ensure rigor and transparency in the review process. The search was based on the principle of comprehensiveness, which is why the Medline and Pubmed databases were used between 2019 and 2024, in English and Portuguese.

The following indexes were used to search for the articles: autism; child; early diagnosis; treatment; quality of life; early diagnosis. The search strategy combined the terms using the Boolean operator "AND" to link the different domains of interest and "OR" to add synonyms or similar words.

Original studies, systematic reviews and meta-analyses were included which discuss the methods, assessment tools and diagnostic criteria used for the early diagnosis of autism in children, as well as approaches related to the effectiveness and accuracy of these methods.

Studies that focused on therapeutic interventions or genetic factors associated with autism were excluded, unless they were directly related to the diagnostic criteria. In addition, studies that did not present specific data on early diagnosis of autism, opinion articles, letters to the editor, conference abstracts, studies that were not available in full text, articles not directly related to diagnostic methods, and those that did not address pediatric populations were excluded.

The evaluation of the data obtained from the articles was divided into two distinct phases: the first, of a descriptive nature, covered the analysis of the period in which the articles were published, the journals with the most publications, among other relevant aspects. The second phase of data analysis consisted of synthesizing, comparing and discussing the information extracted from the articles selected for this study, providing answers to the central research question. Articles requiring a better understanding of the items to be described and analyzed were peer-reviewed.

RESULTS

In the first analysis, the search terms were: “autism”, “child”, “early diagnosis”, “treatment”, “quality of life”, which identified 1,155 articles, 517 of them in Medline and 638 in PubMed. Next, 350 repeated articles and 375 studies that did not deal directly with the defined topic of interest were excluded. In this review, the search uncovered 375 articles related to the subject of this study, 210 of them in Medline and 165 in PubMed. Only 80 articles met the pre-specified eligibility criteria, representing 47 from Medline and 33 from PubMed after reading the introduction and conclusion of the articles. After reading the articles in full, 29 were selected for this review. Finally, 24 articles were used to discuss the work and the others were used for the necessary explanations and descriptions.

The search and selection process was described in the flowchart shown in Figure 1.

Early diagnosis of Autism Spectrum Disorder (ASD) has been widely documented as a determining factor in improving quality of life and child development. An epidemiological surveillance study [1] identified that the prevalence of ASD among 8-year-olds remains stable, but there are large variations in early detection, depending on sociodemographic factors and access to health care.

Another study presented a systematic review that reinforces the substantial benefits of early diagnosis, highlighting that interventions carried out at an early stage allow for the development of more robust communicative and social skills [2]. Similarly, authors argue that behavioral and developmental interventions applied early can significantly minimize the challenges faced by children with ASD throughout their lives [3].

Recent studies also indicate the importance of the family environment as a predictive factor in the development of children with ASD, influencing the effectiveness of interventions [5]. In addition, studies corroborate this idea by discussing how family functioning and emotional aspects can directly impact the results of interventions in children diagnosed early [6].

In addition, intervention models show that a structured approach to early diagnosis can provide a clearer path for the insertion of effective therapeutic interventions, leading to improvements in both the child's behavior and cognitive development [7].

Finally, in a validation study, they reinforce the importance of eliminating the stigma associated with ASD in order to improve the early diagnosis process and ensure that families and health professionals collaborate on more personalized interventions, contributing to a better quality of life [8]. These results have been analyzed in detail in Table 1.

DISCUSSION

INTRODUCTION TO AUTISM

According to the World Health Organization (WHO), Autism Spectrum Disorder (ASD) is a set of conditions characterized by significant challenges in communication and social interaction, as well as atypical behavioral patterns and activities. The manifestations of ASD vary widely and can include anything from speech delays and motor difficulties to

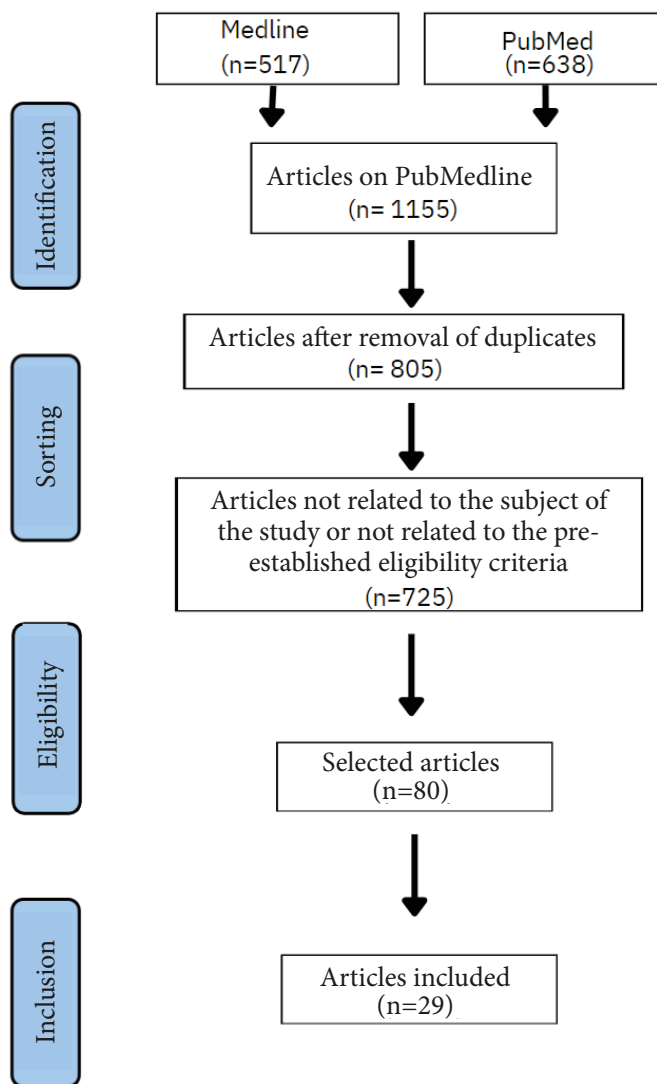


Figure 1. Flowchart of the search and selection of articles. Source: Author

| Title | Author(s) | Type of Study | Objective | Conclusion |
|--|--|------------------------------------|--|--|
| Prevalence and characteristics of autism spectrum disorder among children aged 8 years-Autism and Developmental Disabilities Monitoring Network, 11 Sites, United States, 2018 | Maenner, M. J., Shaw, K. A., Bakian, A. V., Bilder, D. A., et al. 2021 | Epidemiological surveillance study | To analyze the prevalence and characteristics of Autism Spectrum Disorder (ASD) among 8-year-olds in the United States, based on 2018 data obtained by the Autism and Developmental Disabilities Monitoring Network. | The prevalence of ASD among 8-year-olds in the USA was 1 in 54, with variations between different geographical regions. |
| Early Diagnosis of Autism Spectrum Disorder: A Review and Analysis of the Risks and Benefits | Okoye C, Obialo-Ibeawuchi CM, Obajeun OA, Sarwar S, et al. 2023 | Systematic review | This review analyzes the risks and benefits of early diagnosis of ASD, seeking to identify the advantages of early diagnosis to improve development and intervention in children. | Early diagnosis of ASD brings significant benefits by improving interventions, but there are challenges in implementing comprehensive screening. |
| Early intensive behavioral and developmental intervention programs for children with ASD: a literature review | Mota ACW, Vieira ML, Nuerberg AH 2020 | Literature review | The study reviews intensive behavioral and developmental intervention programs for children with ASD, addressing the effectiveness of these interventions on child development. | Behavioral interventions and early intensive development are effective in improving social and cognitive skills in children with ASD. |

| | | | | |
|--|--|-------------------------------------|---|---|
| Identifying, evaluating, and managing children with autism spectrum disorder | Hyman, S. L., Levy, S. E., & Myers, S. M. 2020 | Clinical guide | To provide guidelines for the identification, assessment and management of children with ASD, highlighting the best diagnostic methods and the most effective therapeutic approaches. | It addresses the identification, assessment and management of children with ASD, emphasizing the importance of early intervention. |
| Barriers to autism screening in family medicine practice: a qualitative study | Fenikilé TS, Ellerbeck K, Filippi MK, Daley CM 2015 | Qualitative study | This qualitative study examines the barriers to ASD screening in family medicine practice, identifying the main challenges faced by doctors in the early diagnosis process. | Family doctors face barriers such as lack of time and resources to screen for ASD, which limits early diagnosis. |
| Autism intervention meta-analysis of early childhood studies (Project AIM): updated systematic review and secondary analysis | Sandbank M, Bottema-Beutel K, Crowley LaPoint S, et al. 2023 | Systematic review and meta-analysis | This meta-analysis reviews early interventions for children with ASD, seeking to understand the effectiveness of therapeutic approaches and provide an up-to-date analysis of best practices. | Early interventions for children with ASD have modest effects on cognitive and social development. |
| Sociocultural factors associated with detection of autism among culturally and linguistically diverse communities in Australia | Hussain A, John JR, Dissanayake C, et al. 2023 | Quantitative study | The aim is to investigate the socio-cultural factors that influence the detection of ASD in culturally and linguistically diverse communities in Australia, highlighting the difficulties in accessing diagnosis. | Socio-cultural factors make early detection of ASD difficult in culturally and linguistically diverse communities. |
| Influence of Community-Level Cultural Beliefs about Autism on Families' and Professionals' Care for Children | Kang-Yi CD, Grinker RR, Beidas R, Agha A, et al. 2018 | Qualitative study | It examines how cultural beliefs at the community level affect the way families and professionals care for children with ASD, exploring the relationship between culture and care practices. | Cultural beliefs impact on decisions about the care of children with ASD by both professionals and families. |
| Developmental and Family Environment Predictors of Diagnostic Decision-Making in Children with Autism Spectrum Disorder | Reder M., & Brzezewska, K. 2022 | Longitudinal Study | This study investigates predictors of family environment and development that influence diagnostic decision-making in children with ASD and comorbidities. | The family environment and developmental factors play a crucial role in diagnostic decisions in children with ASD and comorbidities, influencing the accuracy of the diagnosis. |
| Validation of the Greek version of the Affiliate Stigma Scale among mothers of children with autism spectrum disorder | Papadopoulos A, Tafiadis D, Tsapara A, et al. 2022 | Validation Study | The aim is to validate the Greek version of the Affiliate Stigma Scale among mothers of children with ASD, focusing on the impact of affiliate stigma on family well-being. | The Greek version of the Stigma Scale Afiliat was successfully validated among mothers of children with ASD, highlighting the importance of psychological support for these mothers. |
| Early Diagnosis of Autism Spectrum Disorder: A Review and Analysis of the Risks and Benefits | Okoye C, Obialo-Ibeawuchi CM, Obajeun OA, et al. 2023 | Review and Critical Analysis | This article reviews the risks and benefits of early diagnosis of ASD, highlighting the advantages of early intervention and the challenges such as false positives and parental anxiety, seeking a balanced understanding of its importance for child development. | The review highlighted the benefits of early diagnosis for autism, but also highlighted the challenges and risks involved in diagnosis at very young ages. |
| Autism in Brazil: "Cases have not increased, what has increased is the volume of information leading to the diagnosis" | BRAZIL 61 2024 | Expert interview | It reports an expert's perception of the increase in ASD diagnoses in Brazil, attributing the growth to the increase in information available, rather than an increase in cases per se. | The number of autism diagnoses in Brazil has increased due to the greater volume of information available, not necessarily a real increase in cases. |
| DSM-5 TR and ICD-11 - Diagnosis of Autism Spectrum Disorder | Brazil Inclusion Institute 2024 | Informative article | The document addresses the diagnostic guidelines for ASD according to the DSM-5-TR and ICD-11, highlighting the changes and the updated diagnostic criteria. | The article explains the changes in the classifications of autism in the DSM-5 TR and the ICD-11, addressing the implications for the diagnosis and proper care of patients with ASD. |

| | | | | |
|---|---|--|--|---|
| Correio da Saúde - Issue no. 1212 of 12/04/2023 | Paraná Public Prosecutor's Office 2023 | Informative article | The edition of "Correio da Saúde" reports on the prevalence of ASD in Paraná, highlights the importance of raising awareness and reinforces the need for effective public policies for the diagnosis, treatment and social inclusion of people with autism. | It highlights that a growing proportion of the population is diagnosed with ASD, emphasizing the importance of public policies to improve diagnosis and treatment in Brazil. |
| Assessment and care for people with Autism Spectrum Disorders (ASD) | Secretary of Health of Paraná 2023 | Guidance Manual | The document deals with the assessment and care of people with ASD in Paraná, presenting guidelines for diagnosis and care. | It presents guidelines and recommendations for the assessment and care of people with ASD, with a focus on improving early diagnosis and comprehensive treatment. |
| Modified Questionnaire for Autism Screening in Children between 16 and 30 months, Revised, with Follow-up Interview | Robins, D. L.; Fein, D.; Barton, M. 2018 | Screening Tool | It presents the Modified Questionnaire for Autism Screening in Children between 16 and 30 months (M-CHAT-R/F), used for early screening of ASD. | A widely used tool for screening young children for ASD, identifying early signs for appropriate referral. |
| Early diagnosis of Autism Spectrum Disorder is the subject of a new document from the DC for Development and Behavior | Brazilian Society of Pediatrics 2024 | Technical Document | It highlights the importance of early diagnosis of ASD, addressing strategies and recommendations for pediatricians. | It highlights the importance of an early diagnosis of ASD, addressing recommended practices for doctors and pediatricians in order to increase early detection. |
| Meta-Analysis on Intervention Effects of Physical Activities on Children and Adolescents with Autism | Huang, J.; Du, C.; Liu, J.; Tan, G. 2020 | Meta-Analysis | This meta-analysis investigates the effects of physical activities on children and adolescents with ASD, seeking to determine the benefits of these activities for development. | Physical activities have positive effects on the motor and social development of children and adolescents with ASD, suggesting that physical intervention can be an essential part of treatment. |
| Autism Spectrum Disorder: Neurodevelopmental Risk Factors, Biological Mechanism, and Precision Therapy | Wang, L.; Wang, B.; Wu, C.; Wang, J.; Sun, M. 2023 | Literature Review | It explores the neurodevelopmental risk factors and biological mechanisms of ASD, proposing approaches to precision therapy. | It discusses the neurodevelopmental risk factors and biological mechanisms associated with ASD, proposing precision therapies as a promising future avenue for treatment. |
| Autism Spectrum Disorder in Early Childhood: The Center for Baby and Child Studies Model | Martins Halpern, C.; Caldeira da Silva, P.; Costa, D., et al. 2021 | Intervention Model Study | It describes the diagnostic and therapeutic intervention model of the Center for Baby and Child Studies, with a focus on early intervention for ASD. | It describes the diagnostic evaluation and therapeutic intervention model of the Center for Baby and Child Studies, focusing on the treatment of ASD from early childhood. |
| Guidelines for the Rehabilitation of People with Autism Spectrum Disorders (ASD) | Ministry of Health - Health Care Secretariat - Department of Strategic Programmatic Actions | Literature Review and Technical Guidelines | The document aims to guide multi-professional teams in the SUS network in the care and rehabilitation of people with Autism Spectrum Disorder (ASD) and their families, promoting comprehensive and humanized care at the various levels of the health care network. | It emphasizes the importance of early detection, specialized clinical diagnosis and interdisciplinary action to ensure comprehensive care. It emphasizes the need for a Singular Therapeutic Project (PTS) adapted to individual needs, promoting the habilitation and rehabilitation of people with ASD. |
| The importance of a multidisciplinary approach in the treatment of children on the autistic spectrum | De Andrade, B, N, P. et al, 2024. | Literature Review | The study aims to highlight the importance of the multidisciplinary approach in the treatment of Autism Spectrum Disorder (ASD), analyzing how different areas of health, such as psychology, speech therapy and occupational therapy, contribute to the development and well-being of patients. | The treatment of ASD should be personalized and multidisciplinary, focusing on collaboration between professionals to improve patients' quality of life and autonomy. |

| | | | | |
|--|--|---|---|--|
| Diagnostic and Statistical Manual of Mental Disorders, 5th Edition, Text Revision (DSM-5-TR) | American Psychiatric Association (APA) | Revisions of Diagnostic Criteria | The DSM-5-TR aims to provide up-to-date, evidence-based diagnostic criteria for mental disorders, reflecting scientific and clinical advances in the field of mental health. | The DSM-5-TR represents an ongoing effort to align clinical practice with the latest scientific evidence, offering diagnostic guidelines that take into account the diversity and complexity of human experiences. |
| Early Identification of Autism Spectrum Disorder: Recommendations for Practice and Research | Zwaigenbaum, L. 2015 | Multidisciplinary panel of clinicians and researchers | The study aims to review and synthesize the scientific evidence on the early signs and symptoms of Autism Spectrum Disorder (ASD) in children up to 24 months of age, in order to inform best clinical practice and guide future research in the area | The research highlights that early identification of ASD is crucial so that children can access specialized, evidence-based interventions, optimizing long-term results. |
| Early Diagnosis of Autism in the Community Is Associated with Marked Improvement in Social Symptoms Within 1-2 Years | Nitzan Gabbay-Dizdar et al. 2022 | Longitudinal study | To investigate the relationship between age at diagnosis of ASD and the evolution of social symptoms, analyzing whether earlier diagnoses are associated with more significant improvements in social symptoms over time. | The results indicated that children diagnosed before the age of 2.5 were three times more likely to have considerable improvements in the social symptoms of ASD compared to those diagnosed later |

Table 1. Description of the articles selected according to: author, year of publication, type of study and results found. Source: Author.

challenges in socializing and interacting with other children. This diversity reinforces the importance of early diagnosis, as recognizing the signs in the first years of life can significantly impact the child's development and allow for appropriate interventions [9].

However, the variability of symptoms and the lack of knowledge about ASD in society and in health services often result in late diagnosis, hindering access to treatment and compromising individuals' quality of life. Brazilian legislation, with Law n. 12.764/2012, establishes guidelines for the social and professional inclusion of people with ASD, including the right to work and support for their insertion into the market. In addition, Law No. 8,213/1991 makes it possible for the hiring of people with ASD to contribute to meeting inclusion quotas in the workplace, expanding opportunities and promoting the development of an active and dignified life for this population.

Prevalence

ASD is a neurological disorder that can manifest itself as a warning sign of neurodevelopment in the first few months of life, and is approximately four times more prevalent in boys. However, data from the Department of Health suggests an increase in the proportion of diagnoses in girls, reaching a ratio of 3.8/1 between boys and girls [10].

In Brazil, the inclusion of autism in the 2020 demographic census (Law No. 13,861/2019) has allowed for more accurate estimates. According to the Brazilian Institute of Geography and Statistics (IBGE), in the early 2000s, around 2 million Brazilians were considered autistic, representing 1% of the population. Of this total, approximately 400,000 to 600,000 were under the age of 20 and between 120,000 and 200,000 were under the age of 5 [11].

The prevalence of ASD has increased substantially in recent decades: in the early 2000s, one in every 500 children was diagnosed with autism; in 2018, this proportion rose to one in every 44; and in 2020, it reached one in every 26 individuals. In a survey conducted by the Centers for Disease Control and Prevention (CDC) in 2020, it was reported that one in 36

American children up to the age of 8 had a diagnosis of ASD. More recent data from the CDC, released in March 2023, estimates that around 5.6 million Brazilians have ASD, according to data from the 2022 Census, reinforcing the significant increase in this neurodevelopmental disorder [12].

Manifestation of symptoms

The diagnosis of Autism Spectrum Disorder (ASD) is clinical and requires a thorough assessment conducted by a multi-professional team. This process includes a detailed observation of the child and essential information provided by parents or caregivers about development and behavior. Given the variability of ASD manifestations and the absence of specific laboratory or imaging tests, a thorough anamnesis and understanding of the patient's history are crucial for an accurate diagnosis.

The Ministry of Health's Guidelines for the Rehabilitation of People with ASD propose stages focused on identifying "qualitative deviations" in development, with an emphasis on social interaction and language. Psychological and speech therapy assessments complement the diagnosis, helping to establish goals for a Singular Therapeutic Project (PTS) adapted to the child's needs, with guidelines for the family and school [13, 14].

The ICD-10 and DSM-V are used to classify ASD. Both assess developmental delays in social interaction, communication and restrictive behavior patterns, considering that symptoms appear before the age of three. The DSM-V, revised in 2022, adopts strict criteria to avoid overdiagnosis, requiring all sub-criteria to be met in order to confirm the diagnosis [15].

The manifestations of ASD are classified into three levels of severity, which guide appropriate support: level 1 requires light support for mild difficulties in social interaction; level 2 requires substantial support due to marked deficits; and level 3 requires very substantial support for severe social impair-

ment. In restrictive and repetitive behaviors, level 1 indicates mild interference; level 2, frequent behaviors that affect functionality; and level 3, severe difficulties with extreme resistance to change. Additional specifiers, such as intellectual and language deficits and medical conditions, are also considered in diagnosis and support planning [16].

These criteria offer a comprehensive framework that guides accurate diagnoses and allows the formulation of personalized therapeutic strategies for each level of ASD severity.

Different methods and tools for early diagnosis

Early diagnosis of ASD is usually initiated by the parents themselves, who notice signs of atypical development and consult the pediatrician. However, a Brazilian study showed that the first concerns, such as a delay in verbal language, lack of response to the name, absence of eye contact and agitation, are usually noticed at around 23.6 months. Despite this, formal diagnosis occurs on average only at the age of 6, showing a significant delay of approximately 36 months between the initial identification of symptoms and definitive diagnosis.

To facilitate diagnosis, the Brazilian Society of Pediatrics (SBP) recommends that pediatricians use the Modified Checklist for Autism in Toddlers (M-CHAT), translated and validated into Portuguese in 2008 (Sociedade Brasileira de Pediatria, 2019). The M-CHAT-R is a rapid screening tool, applied in routine consultations, which helps to identify early signs of autism with high sensitivity. However, it has low specificity, which means that not all children with risk scores are diagnosed with ASD, and the instrument is not suitable for a complete assessment of child development [17].

If the patient scores positive on the M-CHAT-R, the Follow-up Interview should focus on the areas where the child showed difficulties, replacing the "Yes/No" answers with "Pass/Fail" to improve the accuracy of the screening.

If the child continues to score positive at this stage (M-CHAT-R/F), referral for intervention and in-depth assessment is recommended.

ASD care requires a comprehensive approach, with a complete anamnesis, clinical examination, diagnosis and identification of comorbidities. Diagnostic classification and functional assessment are essential for an early and accurate diagnosis. Based on this diagnosis, a Singular Therapeutic Project (PTS) is developed, adapted to the patient's specific needs, promoting an effective intervention to improve the child's quality of life and development [14].

IMPACT OF EARLY DIAGNOSIS ON QUALITY OF LIFE

Early diagnosis of ASD is crucial for the treatment and management of the condition, as it allows for more effective interventions, conducted by a multi-professional team, which have a positive impact on the psychosocial development of child. Early signs, such as difficulties with social skills and repetitive behaviors, usually appear at 18 months and, when detected, allow for interventions in Applied Behavior Analysis (ABA) with Shared Therapeutic Planning (PTC), aiming for better long-term results [18].

However, early diagnosis faces challenges due to the variability of symptoms and comorbidities, such as ADHD, sleep disorders and gastrointestinal problems. Studies indicate that early diagnosis (12-24 months) results in fewer developmental delays compared to late diagnosis (25-41 months), although all children with ASD exhibit deficits in language and social communication [19].

Family participation is fundamental, as informed caregivers can speed up diagnosis and facilitate treatment by providing accurate information about the child. However, the diagnosis of ASD requires more intense involvement from parents, who often report high levels of stress and stigmatization, affecting

mental health and the family's quality of life [5, 8]. Thus, early diagnosis of ASD benefits not only child development, promoting advances in communication and social skills, but also the general well-being of the family, reducing stress and long-term costs with intensive therapies and specialized educational services [2].

BARRIERS TO EARLY DIAGNOSIS AND TREATMENT MODELS

As has been shown, early diagnosis of ASD is essential in order to implement interventions that promote child development and increase children's quality of life. However, several barriers hinder this identification, such as the wide variation of symptoms, the lack of specialized training among health professionals and educators, and the stigma that still surrounds ASD. These issues can delay the recognition of early signs and thus the start of interventions [6, 18, 20]. Socio-economic, cultural and geographical factors also affect access to early diagnosis: low-income families and those living in rural areas have less access to specialist consultations, while some cultures, due to misinformation or prejudice, still avoid seeking help for these cases [21, 22].

To overcome these challenges, cooperation between health professionals, educators, families and public policies is crucial, as are investments in training and awareness. Early interventions, including music therapy, cognitive-behavioral and social-behavioral therapies, offer important advances in social and emotional skills. Therapies such as brain stimulation, vitamin supplementation and dietary adjustments (gluten and casein free) have also shown benefits in gastrointestinal and behavioral problems, although more studies are needed for validation [23, 24]. In pharmacological treatment, drugs such as aripiprazole and risperidone help to control aggressive and repetitive behaviors.

More recent therapeutic approaches, such as stem cell therapy, have shown potential in reducing inflammation and improving the well-being of ASD patients. In Brazil, specialized centers, such as the Center for Baby and Child Studies, offer comprehensive support with speech, occupational and psychological therapies and intensive care, promoting a holistic and personalized approach. This combination of support and varied therapies has shown promising results, favoring children's development and providing a significant improvement in the quality of life for both patients and their families [7]. In summary, early diagnosis and personalized interventions are essential for optimizing the development of children with ASD and effectively supporting their families.

CONCLUSION

Autism Spectrum Disorder (ASD) is a neurological disorder with varied manifestations that requires an individualized approach to diagnosis and treatment. Early detection is

crucial to promote development and quality of life, allowing for specific interventions that help with communication, socialization and behaviour. However, lack of knowledge and the diversity of symptoms often delay diagnosis, hindering access to the necessary care.

In Brazil, legislation such as Law No. 12.764/2012 and Law No. 8.213/1991 guarantee the social and professional inclusion of people with ASD, reinforcing the need for policies that offer support and opportunities. Strategies such as the use of M-CHAT and the Singular Therapeutic Project (PTS) have proved effective in improving diagnosis and interventions, benefiting both the patient and their family.

Thus, advances in early diagnosis and treatment practices, together with inclusive policies, are essential for improving the quality of life of people with ASD. Investments in professional training, social awareness and access to specialized services are key to promoting the integration and well-being of this population.

REFERENCES

- Maenner MJ, Shaw KA, Bakian AV, Bilder DA, Durkin MS, Esler A, et al. Prevalence and characteristics of autism spectrum disorder among children aged 8 years—Autism and Developmental Disabilities Monitoring Network, 11 Sites, United States, 2018. *MMWR Surveill Summ.* 2021;70(11):1 [1].
- Okoye C, Obialo-Ibeawuchi CM, Obajeun OA, Sarwar S, Tawfik C, Waleed MS, et al. Early Diagnosis of Autism Spectrum Disorder: A Review and Analysis of the Risks and Benefits. *Cureus.* 2023;15(8). doi: 10.7759/cureus.43226. PMID: 37692637; PMCID: PMC10491411 [2].
- Mota ACW, Vieira ML, Nuernberg AH. Programas de intervenções comportamentais e de desenvolvimento intensivas precoces para crianças com TEA: uma revisão de literatura. *Rev Educ Espec.* 2020;33/1-27. Disponível em: <https://periodicos.ufsm.br/educacaoespecial/article/view/41167> [3].
- Hyman SL, Levy SE, Myers SM. Identifying, evaluating, and managing children with autism spectrum disorder. *Pediatrics.* 2020;145(1) e20193447 [4].
- Reder M, Brzezewska K. Developmental and family environment predictors of diagnostic decision-making in children with autism spectrum disorder and comorbid disorders. *Health Psychol Rep.* 2022;10(4):280-93. doi: 10.5114/hpr/151836. PMID: 10501433 [5].
- Fenikilé TS, Ellerbeck K, Filippi MK, Daley CM. Barriers to autism screening in family medicine practice: a qualitative study. *Prim Health Care Res Dev.* 2015;16(4):356-66. doi: 10.1017/S1463423614000449. PMID: 25367194 [6].
- Martins Halpern C, Caldeira da Silva P, Costa D, Nascimento MJ, Mesquita Reis J, Martins MT, et al. A Perturbação do Espectro do Autismo na Primeira Infância: O Modelo do Centro de Estudos do Bebê e da Criança de Avaliação Diagnóstica e Intervenção Terapêutica. *Acta Med Port.* 2021;34(10):657-63. doi: 10.20344/amp.13397. PMID: 33507861 [7].
- Papadopoulos A, Tafiadis D, Tsapara A, Skapinakis P, Tzoufi M, Sifaka V. Validation of the Greek version of the Affiliate Stigma Scale among mothers of children with autism spectrum disorder. *BJPsych Open.* 2022;8(1). doi: 10.1192/bjo.2021.1083. PMID: 35045904; PMCID: PMC8811780 [8].

Organização Mundial Da Saúde (OMS). Transtorno do espectro autista (TEA). Genebra: Organização Mundial da Saúde, 2021. Disponível em: <https://www.who.int/pt/news-room/fact-sheets/detail/autism-spectrum-disorders>. Acesso em: 24 fev. 2025 [9].

Secretaria de Saúde do Paraná. Avaliação e Atendimento à pessoa com Transtorno do Espectro do Autismo (TEA). Secretaria de Saúde do Paraná. 2023. Disponível em: https://www.saude.pr.gov.br/sites/default/arquivos_restritos/files/documento/2023-02/1a_edicao.pdf. Acesso em: 21 ago. 2024 [10].

Ministério Público do Paraná. Correio da Saúde - Edição nº 1212 de 12/04/2023. Disponível em: <https://site.mppr.mp.br/saude/Pagina/Correio-da-Saude-Edicao-ndeg-1212-de-12042023#:~:text=Atualmente%2C%20o%20Instituto%20Brasileiro%20de,da%20popula%C3%A7%C3%A3o%20estaria%20no%20espectro>. Acesso em: 21 ago. 2024 [11].

Brasil 61. Autismo no Brasil: “Casos não aumentaram, o que aumentou foi o volume de informações que levam ao diagnóstico” diz especialista. Disponível em: <https://brasil61.com/n/autismo-no-brasil-casos-nao-aumentaram-o-que-aumentou-foi-o-volume-de-informacoes-que-levam-ao-diagnostico-diz-especialista-bras239991#:~:text=N%C3%BAmeros%20no%20Brasil&text=Com%20uma%20popula%C3%A7%C3%A3o%20estimada%20em,crian%C3%A7as%20apresentava%20TEA%20naquele%20ano>. Acesso em: 21 ago. 2024 [12].

Ministério Da Saúde. Diretrizes de Atenção à Reabilitação da Pessoa com TEA. Brasília: Ministério da Saúde. Acesso em: 24 fev. 2025 [13]

De Andrade, B. N. P.; Pereira, G. E. T.; E Dias, G. S.; Silva, G. B. B.; Pereira, G. H. G.; Pereira, J. F. E.; Gonzaga, M. E. C.; Valentim, M. E. Z.; Da Costa, M. E.; MORAES, S. M. A importância do abordagem multidisciplinar no tratamento de crianças com espectro autista. *Brazilian Journal of Health Review*, [S. l.], v. 7, n. 1, p. 3568–3580, 2024. DOI: 10.34119/bjhrv7n1-288. Disponível em: <https://ojs.brazilianjournals.com.br/ojs/index.php/BJHR/article/view/66786>. Acesso em: 24 fev. 2025 [14].

American Psychiatric Association. Diagnostic and statistical manual of mental disorders: DSM-5-TR. Washington, DC: American Psychiatric Publishing, 2022 [15].

Instituto Inclusão Brasil. DSM-5 TR E CID-11 – Diagnóstico de Transtorno do Espectro Autista. Disponível em: <https://institutoinclusaobrasil.com.br/dsm-5-tr-e-cid-11-diagnostico-de-transtorno-do-espectro-autista/>. Acesso em: 21 ago. 2024 [16].

Sociedade Brasileira de Pediatria. Diagnóstico precoce para o Transtorno do Espectro do Autismo é tema de novo documento do DC de Desenvolvimento e Comportamento. Disponível em: <https://www.sbp.com.br/imprensa/detalhe/nid/diagnostico-precoce-para-o-transtorno-do-espectro-do-autismo-e-tema-de-novo-documento-do-dc-de-desenvolvimento-e-comportamento/>. Acesso em: 21 ago. 2024 [17].

Zwaigenbaum, L. et al. Early identification of autism spectrum disorder: Recommendations for practice and research. *Pediatrics*, v. 136, Supplement 1, p. S10–S40, 2015 [18].

Gabbay-Dizdar, N., Ilan, M., Meiri, G., Faroy, M., Michaelovski, A., Flusser, H., Menashe, I., Koller, J., Zachor, D. A., & Dinstein, I. (2022). Early diagnosis of autism in the community is associated with marked improvement in social symptoms within 1–2 years. *Autism*, 26(6), 1353–1363. <https://doi.org/10.1177/13623613211049011> [19].

Sandbank M, Bottema-Beutel K, Crowley LaPoint S, Feldman JI, Barrett DJ, Caldwell N, et al. Autism intervention meta-analysis of early childhood studies (Project AIM): updated systematic review and secondary analysis. *BMJ*. 2023;383. doi: 10.1136/bmj-2023-076733. PMID: 37963634; PMCID: PMC10644209 [20].

Hussain A, John JR, Dissanayake C, Frost G, Girdler S, Karlov L, et al. Sociocultural factors associated with detection of autism among culturally and linguistically diverse communities in Australia. *BMC Pediatr*. 2023;23(1):415. doi: 10.1186/s12887-023-04236-2. PMID: 37612588; PMCID: PMC10463473 [21].

Kang-Yi CD, Grinker RR, Beidas R, Agha A, Russell R, Shah SB, et al. Influence of Community-Level Cultural Beliefs about Autism on Families' and Professionals' Care for Children. *Transcult Psychiatry*. 2018;55(5):623–47. doi: 10.1177/1363461518779831. PMID: 29972327; PMCID: PMC7008392 [22].

Wang L, Wang B, Wu C, Wang J, Sun M. Autism Spectrum Disorder: Neurodevelopmental Risk Factors, Biological Mechanism, and Precision Therapy. *Int J Mol Sci*. 2023;24(3):1819. doi: 10.3390/ijms24031819. PMID: 36768153; PMCID: PMC9915249 [23].

Huang J, Du C, Liu J, Tan G. Meta-Analysis on Intervention Effects of Physical Activities on Children and Adolescents with Autism. *Int J Environ Res Public Health*. 2020;17(6):1950. doi: 10.3390/ijerph17061950. PMID: 32192008; PMCID: PMC7142971 [24].