



## CRITICAL ANALYSIS OF THE GROWTH OF AUTISM SPECTRUM DISORDER DIAGNOSES IN THE LIGHT OF SCIENTIFIC ADVANCES: PROGRESS IN THE IDENTIFICATION OR INCREASE IN PREVALENCE?



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## ABSTRACT

Autism Spectrum Disorder (ASD) has stood out in recent decades for a significant growth in the number of diagnoses at a global level. This phenomenon raises questions about whether this increase reflects a real growth in the prevalence of the condition or whether it is mostly related to the evolution of diagnostic criteria and the improvement of clinical identification methods. This study aims to investigate the extent to which scientific, methodological and social advances have influenced the diagnostic rates of ASD, to the detriment of an effective increase in incidence. This is an integrative literature review, with a qualitative approach and exploratory character. Scientific articles, guidelines, and official documents published between 2000 and 2024, available in databases such as PubMed, Scopus, SciELO, Lilacs, and Web of Science, were analyzed. The data were analyzed in the light of the transformations observed in diagnostic manuals, such as the DSM-5, as well as international epidemiological registries, such as those of the Centers for Disease Control and Prevention (CDC). The results indicate that significant changes in the classification criteria, greater training of health professionals, increased access to assessment services, in addition to increased awareness of society and families about the disorder, are determining factors for the increase in the number of diagnoses. In addition, the inclusion of more subtle manifestations of the spectrum is highlighted, such as mild and atypical cases, which previously went unnoticed. Although the role of environmental and epigenetic factors in the increase in prevalence cannot be completely ruled out, the main factor responsible for the increase in ASD diagnoses is related to technical-scientific advances and the greater sensitivity of health systems to the early and accurate identification of the disorder.

**Keywords:** Autism Spectrum Disorder. Diagnosis. Diagnostic Criteria. Prevalence. Neurodevelopment.

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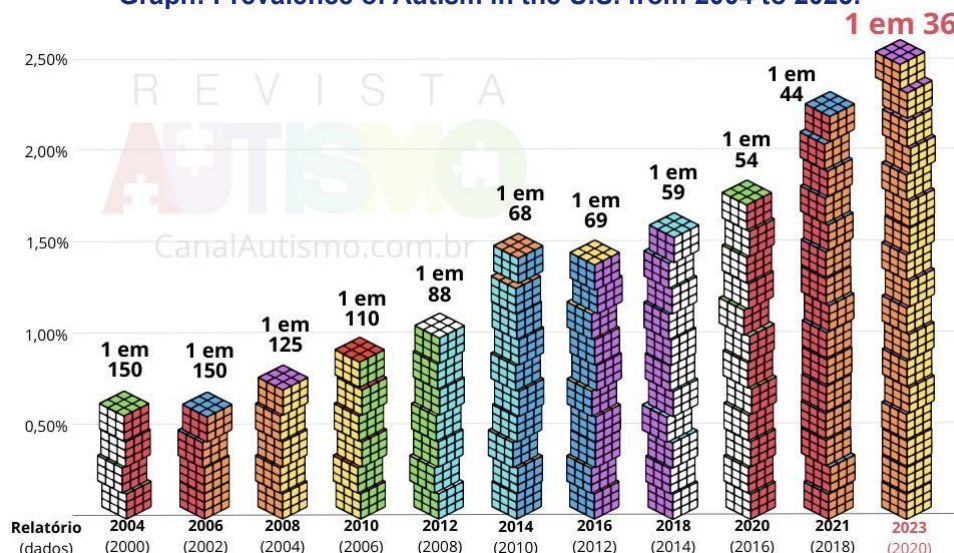
## INTRODUCTION

Autism Spectrum Disorder (ASD) is a neurodevelopmental condition characterized by difficulties in communication and social interaction, as well as restricted and repetitive patterns of behavior (AMERICAN PSYCHIATRIC ASSOCIATION, 2014). ASD presents itself in a diverse way, with significant variations in level of support, cognition and language, and is therefore classified as a spectrum. Recent data reveal a significant growth in the prevalence of ASD diagnoses in the world. The Centers for Disease Control and Prevention (CDC), in the United States, updated its estimates in 2023, pointing out that 1 in 31 children is on the spectrum (CDC, 2023). Previously, this ratio was 1 in 36 (CDC, 2020).

The CDC is a North American agency focused on public health, which monitors and investigates health conditions, being one of the world references in epidemiology. In Brazil, according to data from the Ministry of Health, there are still gaps in national mapping, making it difficult to obtain accurate data. However, ongoing studies, such as those conducted by researchers from the University of São Paulo (USP), suggest that the prevalence may be even higher than that pointed out by international studies.

These studies have not yet been officially published, but they point to an underreporting of cases and late diagnoses. Thus, ASD presents itself not only as a clinical challenge, but also as a public health issue. The increase in diagnoses raises questions about the real prevalence of the condition, requiring a critical analysis of the evolution of diagnostic criteria, the training of qualified professionals and access to health services. In addition, ASD affects boys and girls differently. Studies indicate that boys are diagnosed more frequently, in a ratio of approximately 4:1 (LOMBROSO; GESCHWIND, 2006), which may reflect biases in diagnostic instruments that do not sufficiently contemplate the typical behavioral profiles of the female sex. It is also essential to discuss the ableism faced by autistic people, in addition to the legal and structural gaps that affect access to diagnosis, education, social inclusion, and specialized care.

**Graph: Prevalence of Autism in the U.S. from 2004 to 2023.**



**Source:** Centers for Disease Control and Prevention (CDC), 2023. Adapted from Autism Channel

Furthermore, as mentioned above, ableism is any form of prejudice, discrimination or exclusion directed at people with disabilities, whether physical, sensory, intellectual, mental or neurodivergent (as in the case of people with autism). This prejudice is based on the idea that people with disabilities are inferior, less capable, or less worthy of respect and opportunities. This behavior can manifest itself in various ways, for example: Jokes or offensive comments about the disability; Lack of accessibility (physical, communicational, educational); Denying job or education opportunities based on the person's condition; Treating the person with disabilities as a "poor thing" or "inspiring" just for existing; Ignoring the autonomy and decisions\*\* of these people. Ableism can be both explicit (aggressions, direct exclusions) and subtle (microaggressions, paternalistic or stereotyped attitudes). In Brazil, ableism is fought by laws that ensure the rights of people with disabilities, including those with Autism Spectrum Disorder (ASD). Ableism is defined as any form of discrimination, prejudice, or exclusion directed at people with disabilities. The main legislations that deal with this are:

1. Brazilian Law for the Inclusion of Persons with Disabilities (Law No. 13,146/2015). Also called the Statute of Persons with Disabilities, this law prohibits ableism and guarantees equal opportunities.

Article 4: "Every person with disabilities has the right to equal opportunities with other people and will not suffer any kind of discrimination.

"Article 88: "It is a crime punishable by imprisonment and a fine to practice, induce or incite discrimination against a person on the basis of his or her disability."

2. Law No. 12,764/2012 – National Policy for the Protection of the Rights of Persons with Autism Spectrum Disorder. It recognizes the person with ASD as a person with disabilities, guaranteeing them all the corresponding rights, including protection against ableism.

Article 1, § 2: "The person with autism spectrum disorder is considered a person with a disability, for all legal purposes."

Art. 7: Provides punishments for any form of discrimination against people with ASD.

Autism Spectrum Disorder (ASD) is multifactorial and complex, involving interactions between genetic, epigenetic, environmental, immunological, and neurobiological factors. Several studies point to alterations in genes related to synaptic development and functioning, such as MECP2, SHANK3, and NRXN1, as well as epigenetic modifications that affect gene expression during neurodevelopment. In the brains of individuals with ASD, altered patterns of neural connectivity are observed, with local hyperconnectivity and long-distance hypoconnectivity, especially in regions such as the amygdala, the prefrontal cortex, the cerebellum, and the hippocampus. This imbalance affects the integration of sensory, social, and behavioral information. In addition, there is evidence of dysfunction in neurotransmission, with emphasis on the imbalance between the glutamatergic and GABAergic systems, as well as alterations in the dopaminergic, serotonergic, and oxytocinergic systems, which contribute to the characteristic symptoms of ASD. The immune system also shows significant changes, such as chronic microglial activation and increased pro-inflammatory cytokines, suggesting a state of neuroinflammation. This condition can be influenced by maternal infections during pregnancy and dysfunctions of the blood-brain barrier. Additionally, there is a growing relationship between gut dysbiosis and ASD symptoms, reinforcing the importance of the gut-brain axis in modulating behavior. Alteration of the gut microbiota can lead to the production of neuroactive metabolites, interfering with the central nervous system through neural, hormonal, and immune pathways. Oxidative stress and mitochondrial dysfunctions are also described, with impaired energy production and increased reactive oxygen species, aggravating neurological damage during development. Together, these factors contribute to the establishment of a heterogeneous neurobiological profile that is still under investigation, characteristic of ASD.

The growing incidence of ASD diagnoses in recent decades has generated debate in the scientific, clinical, and social circles. There is an urgent need to differentiate whether this increase is a consequence of a real growth in prevalence or if it only reflects advances in diagnostic instruments, greater professional training and expansion of social knowledge about the spectrum. Thus, this work is justified by the importance of understanding what is behind the increase in the number of diagnosed cases of ASD, allowing more assertive public actions, improving the health network and combating misinformation.

Thus, the objective of this research was to investigate to what extent the increase in the number of diagnoses of autism spectrum disorder reflects advances in identification methods and diagnostic criteria, rather than an effective growth in the prevalence of the condition.



## METHODOLOGY

This is a qualitative and exploratory study, with a design based on an integrative literature review. The research was conducted with the aim of investigating whether the increase in the number of diagnoses of Autism Spectrum Disorder (ASD) in recent decades is predominantly due to advances in identification methods, greater social awareness and expansion of diagnostic criteria, as established in classification manuals such as the DSM and ICD, or if there is evidence to support a real growth in the prevalence of the condition. Data collection was carried out through a systematized search in national and international databases, including PubMed, Scopus, SciELO, Web of Science, Lilacs, and Google Scholar, with articles published between 2000 and 2024. Controlled and uncontrolled descriptors were used, such as: "autism spectrum disorder", "diagnosis", "diagnostic criteria", "prevalence", "early identification", "neurodevelopment" and their English counterparts.

The Boolean operators AND and OR were applied to refine the results. The selection of studies followed the inclusion criteria: original articles, systematic reviews, diagnostic guidelines, and official documents that addressed the growth of ASD diagnoses and changes in identification criteria. Duplicate articles, studies with low methodological quality or that were not directly related to the theme were excluded. Data analysis was carried out through critical reading and thematic categorization of the selected studies, allowing the construction of an integrative synthesis. To ensure the quality and validity of the data, the PRISMA protocol (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) was followed, adapted to the proposal of an integrative review. The results obtained were discussed in the light of the current literature and compared with the changes that occurred in the editions of the DSM and the ICD, as well as with the epidemiological records of reference institutions such as the CDC (Centers for Disease Control and Prevention) and the World Health Organization (WHO).

## RESULTS

The literature review revealed that the exponential growth in the diagnoses of Autism Spectrum Disorder (ASD) is not linked to a proportional increase in prevalence in the general population, but rather to the transformation of the clinical and social view of neurodevelopment. One of the most relevant findings concerns the expansion of diagnostic boundaries, which allowed the inclusion of profiles previously considered "different", "special", "weird", "isolated", but later legally considered the term deficient. It was found that, with the advancement of neuroscience and child psychiatry, autism is no longer

restricted to severe conditions with intellectual delay or total absence of language. Currently, individuals with preserved cognitive functioning, developed verbal skills, and social compensation strategies are now correctly included in the spectrum, evidencing a new clinical and epidemiological panorama.

Another result observed was the direct impact of changes in the diagnostic manual (DSM), especially the transition from DSM-IV to DSM-5. The replacement of subcategories such as classic autism, Asperger's syndrome, and disintegrative disorder with a single spectrum diagnosis increased the sensitivity for identifying varied manifestations, although it also generated confusion and overlap with other neuropsychiatric conditions. Based on the data analyzed, a correlation was also identified between the increase in diagnoses and the greater availability of trained professionals. In previous years, the lack of neuropsychiatrists, child psychiatrists, and trained psychologists made it difficult to recognize the condition early, especially in peripheral regions. With the growth of the mental health care network, more screening centers and screening protocols have emerged that have facilitated access to diagnosis.

It was also found that the educational and family context decisively influenced the mapping of cases. The increase in children's schooling and the demand for social adaptation in educational institutions favored the referral of children for neurological and psychosocial evaluation. Many behaviors that were previously normalized or attributed to "shyness", "difficult temper" or "upbringing" have come to be reinterpreted from the perspective of neurodiversity. Another relevant point was the finding that the current criteria still do not adequately contemplate autistic traits in girls and in diverse populations. The literature pointed to a tendency towards female invisibility, since many girls are able to mask the signs for longer, making early diagnosis difficult and contributing to a gap in the statistics. Finally, it was identified that the trivialization of the term "autism" in public discourse and on social networks generated ambiguous effects: while contributing to destigmatizing the condition, it also favored hasty diagnoses, unconfirmed self-diagnoses, and a superficial understanding of the complexity of the spectrum.

## **DISCUSSION**

### **THE LEVELS OF THE AUTISM SPECTRUM**

According to the Diagnostic and Statistical Manual of Mental Disorders – 5th edition (DSM-5) in force in Brazil, ASD is classified into three levels of support, according to the individual's need for support (AMERICAN PSYCHIATRIC ASSOCIATION, 2014):

1. Level 1: Requires support. Individuals with some functional independence but significant social difficulties.
2. Level 2: Requires substantial support. More evident difficulties in verbal and non-verbal communication, with accentuated repetitive behaviors.
3. Level 3: Requires very substantial support. Individuals with great difficulty in communication, often non-verbal, with very restrictive behaviors.

Despite this categorization, several studies point to the limitation of these criteria in representing the diversity of clinical manifestations (LEEKAM et al., 2011). Many autistic people have characteristics that oscillate between levels, which makes diagnosis a subjective process and dependent on the clinical sensitivity of the evaluator.

## GENETIC AND ENVIRONMENTAL FACTORS: A POLYGENIC VIEW

ASD has a significant genetic component, but not determined by a single gene. Studies point to a complex and polygenic genetic architecture, with hundreds of genes involved (SANDIN et al., 2014). In addition, de novo mutations and alterations in neurodevelopmental-related genes, such as CHD8, SHANK3 and MECP2, have been increasingly studied (TAMMIMIES et al., 2015). Environmental factors also play a relevant role, such as exposure to pollutants, viral or bacterial infections during pregnancy, advanced paternal age, and substance use (VARGAS et al., 2020). Epigenetics emerges as a bridge between these factors, explaining how the environment can modulate gene expression without altering the DNA sequence (NGUYEN et al., 2010).

## THE TRIVIALIZATION OF DIAGNOSIS

With the increase in awareness about ASD, many individuals have started to seek specialized evaluation. However, a concern arises: the trivialization of the diagnosis. Rapid diagnoses without well-established criteria have become common, especially in school contexts or in non-specialized care. Trivialization can invalidate real experiences of autistic individuals who need support (CAMPBELL, 2009). In addition, it can overload public policies and health services, diverting resources and commitments.

## DIAGNOSTIC ADVANCES AND QUALIFIED PROFESSIONALS

Another factor to be considered in the increase in diagnoses is the growth of professional training. In recent decades, there has been a significant expansion in the training of doctors, psychologists, psychiatrists, neuropsychopedagogues and therapists



specialized in neurodevelopment. More trained professionals who are sensitive to spectrum signals contribute to earlier and more accurate identification (OLIVEIRA; FERREIRA; COSTA, 2021).

This advance is also related to the expansion of assessment instruments, such as ADOS-2 (Autism Diagnostic Observation Schedule) and ADI-R (Autism Diagnostic Interview – Revised), which are applied by multiprofessional teams in a controlled environment.

### DIAGNOSTIC CRITERIA: LIMITATIONS OF DSM-5

Despite its wide use, DSM-5 has limitations. The dependence on criteria such as difficulties in social communication and repetitive behaviors can exclude individuals with preserved intelligence, masking strategies or atypical profiles, especially in girls (LUGNEGÅRD et al., 2012). Often, the clinical view is decisive for the diagnosis, even if the standardized criteria are not completely met.

It is important to note that the term "disease", present in the name of the CDC (Centers for Disease Control and Prevention). ASD is not a disease, but a neurodevelopmental condition. The CDC's nomenclature refers to terminology in the United States, but it can imply the pathology of autism itself.

### GENDER RELATIONS AND FEMALE INVISIBILITY

Girls are often underdiagnosed or diagnosed later. They tend to have more internalized behaviors and more developed social imitation skills, which masks typical signs of the spectrum (KIRKOVICZ et al., 2019). This gender bias contributes to the underestimation of the real prevalence of ASD among women.

Image: Prevalence of Autism by Sex



Source: CDC, 2021.

## ABLEISM AND SOCIAL BARRIERS

Autistic people face ableism in various spheres — from education to the job market. Social marginalization is aggravated by the lack of effective public policies and the social misunderstanding of individual needs. According to Law No. 12,764/2012, which institutes the National Policy for the Protection of the Rights of Persons with Autism Spectrum Disorder, ASD is recognized as a disability for legal purposes in Brazil, guaranteeing these people the rights provided for in Brazilian legislation.

## CONCLUSION

The increase in diagnoses of Autism Spectrum Disorder in recent decades is a multifactorial phenomenon. This growth cannot be attributed solely to the real increase in the prevalence of the condition in the population, but to a set of factors such as the expansion of diagnostic criteria, greater clinical sensitivity, scientific advances, and an increase in the number of professionals qualified to evaluate and monitor people on the spectrum. The evolution of science has allowed the understanding of ASD as a spectrum with different levels of support and heterogeneous manifestations.

Genetics, in turn, demonstrated that the disorder has a polygenic character, strongly influenced by environmental, epigenetic and contextual factors. The reductionist and pathologizing view, which previously restricted diagnosis to severe conditions, gives way to an approach that is more inclusive and sensitive to human neurobiological diversity. However, there are still many challenges: the trivialization of diagnosis, the limiting criteria of the DSM-5, the invisibility of autistic women, and structural ableism. These points require critical vigilance and constant updating of the clinical, legal and social instruments that involve ASD. It is essential to remember that autism is not a disease, but a neurological condition that demands understanding, inclusion, and respect for differences. The fight for effective public policies, access to early diagnosis and adequate treatment, as well as for breaking social stigmas, must be permanent.

Brazilian science has also been committed to contributing with relevant data, despite the lack of large population studies and the need for greater investment in research. It is expected that, in the coming years, these gaps will be filled and national epidemiological data will be able to more accurately reflect the Brazilian reality. Based on the analysis developed, it is concluded that the increase in the number of ASD diagnoses mostly reflects advances in identification methods, social awareness and changes in clinical criteria, rather than an effective growth of the condition itself. Therefore, a critical reading of the data is



essential to avoid misinterpretations and to promote truly humanized and scientific care for autistic people and their families.

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