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Autism Spectrum Disorder: The Remarkable Gap Between Diagnostic And Effective Care

Meriem Touati[1], Lounes Lallem[2], Fatma Imene Kaour[3], Lamia Ouldmohand[4]

[1]Associate-professor, higher school for teachers of deaf and mute, (Algeria).
[2]Associate-professor, Faculty of human and social sciences, University of Tizi Ouzou, Multidisciplinary laboratory of Human Sciences, Environment and Society, (Algeria).
[3]Associate-professor, Faculty of human and social sciences, University of Annaba, laboratory for work analysis and ergonomic studies, (Algeria).
[4]Associate-professor, Faculty of human and social sciences, University of Bouira, Multidisciplinary laboratory of Human Sciences, Environment and Society, (Algeria)
The Author's E-mail: Meriem.touati@ummto.dz[1], lounes.lallem@ummto.dz[2], imene.kaour23@gmail.com[3], l.ouldmohand@univ-bouira.dz[4]

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Abstract:
Autism spectrum disorder (ASD) is a neurodevelopmental disorder that manifests early in childhood, significantly affecting cognitive, social, and emotional functioning. Despite its role in advancing the field of child psychiatry, ASD remains a topic of ongoing debate and discussion. This is true at various levels, including its diagnostic characteristics, epidemiology, causal hypotheses, as well as approaches to rehabilitation and treatment. Although considerable progress has been made, particularly in the fields of neuroscience and biology, controversies persist. In this paper, we aim to explore, through an analysis of the currently available data, the different aspects of the ongoing debates surrounding autism spectrum disorder.

Keywords: Autism spectrum disorder; clinical diagnosis; epidemiological characteristics; causal hypotheses; care challenges.

1. Introduction:

Autism spectrum disorder (ASD) is a neurodevelopmental disorder that appears early in the affected child's life, significantly influencing their cognitive, social, and emotional functioning. Despite being one of the syndromes that have greatly contributed to the foundation of child psychiatry, ASD remains a subject of ongoing debate and controversy across all levels. These debates pertain to its diagnostic characteristics, epidemiology, causal approaches, as well as rehabilitation and treatment strategies. With the intense media coverage of this disorder, the disagreements and controversies have only intensified, revealing various conflicts of a cognitive, ideological, ethical, and even economic nature.

The medical and scientific community has worked to adjust diagnostic criteria, which has led to an increase in prevalence rates. Consequently, new causes have been proposed, and autism has been redefined as a neurobiological disorder. Politically, laws and regulations concerning ASD have been enacted, while economically, markets related to autism spectrum disorder have been created and exploited—ranging from medications, diverse training programs, books of all kinds, special diets, genetic tests, to the promotion of educational and behavioral methods, and various treatments (Chamak, 2021).

All these controversies have affected the lives of affected children, causing their parents to experience confusing and often harsh realities, particularly in the absence of a clear and precise strategy for managing the disorder. The situation is further complicated by a lack of adequately trained specialists in both diagnosis and treatment, as well as the existence of multiple, often ineffective and expensive care mechanisms.

Thus, in this contribution, we aim to explore the various circumstances surrounding autism spectrum disorder, particularly with regard to

diagnostic data, the strategies in place and their effectiveness, as well as the epidemiological characteristics and how they have been addressed. Additionally, we will examine the factors contributing to discrepancies in the data, and the portrayal of ASD as a widespread issue of public health concern. We will also delve into the challenges related to causal hypotheses, alongside the available therapeutic and rehabilitative methods.

2. Defining Autism Spectrum Disorder and the Diagnostic Challenges:

Since its first description by Kanner in 1943, the definition and understanding of autism have evolved significantly. According to the fifth edition of the (Diagnostic and Statistical Manual of Mental Disorders (DSM-5)), ASD is characterized by various impairments in social and emotional communication, alongside restricted interests and repetitive or stereotyped behaviours (APA, 2013). Symptoms must be present in early childhood, though their impact on functioning may not become apparent until later stages of development, such as when social demands exceed the individual's capacities (e.g., upon entering school or during adolescence). For a complete ASD diagnosis, symptoms must significantly affect functioning across different contexts, such as school, family, or professional settings (APA, 2013).

When diagnosing ASD, several characteristics must be clarified. First, it is essential to assess the severity of the disorder, which is classified as "mild," "moderate," or "severe," depending on the individual's need for support. Additionally, according to the DSM-5, the presence of co-occurring conditions, such as intellectual disability, language impairments, or other medical, genetic, or developmental disorders (e.g., catatonia), must also be noted (APA, 2013).

ASD is an extremely complex disorder, with its symptoms influenced by individual characteristics like gender, age, comorbidities, and culture. Its complexity is further compounded by the lack of clear biological markers, meaning that diagnosis is based exclusively on behavioural observations, which can vary significantly from one child to another and even within the same child across different stages of development. This variability gives rise to other diagnostic challenges, particularly concerning differential diagnosis and comorbid disorders, as well as early diagnosis.

In the absence of clinical evidence, differential diagnosis relies on purely clinical elements. Delays in developmental milestones, language impairments, social isolation, and various social interaction difficulties—as well as certain strange or stereotyped behaviours—are all factors that can overlap and complicate the diagnostic process (Dormoy, 2020). Various life difficulties can lead to periods of delayed reciprocal communication, disrupted or altered symbolic expression, or even its total breakdown, which may be caused by factors unrelated to autism. For instance, children suffering from neglect, family instability, or deprivation may exhibit behaviours resembling those of autism, such as repetitive and stereotyped behaviours or abnormal sensory responses. Additionally, certain sensory integration disorders may create a false impression of an autism-like syndrome. It is also not always easy to distinguish between obsessive-compulsive disorder (OCD) and Asperger's syndrome. Severe hyperactivity might lead to a temporary misdiagnosis. Furthermore, some young children may experience unexplained developmental stagnation (Lemay, 2020). Other disorders, such as social anxiety, early-onset schizophrenia, sensory impairments like deafness, and Rett syndrome, may have overlapping symptoms with ASD, making differential diagnosis particularly challenging (Bourseau et al., 2020).

Another issue lies in the difficulty of early ASD diagnosis. Most researchers agree on the importance of early diagnosis, as the first years of life are a period of significant brain plasticity, during which therapeutic interventions can be highly effective in achieving optimal treatment outcomes. However, the challenge of early diagnosis involves addressing key questions: At what age can we expect to diagnose autism in a child? What are the signs? And is autism diagnosis sensitive enough before 24 months of age? In reality, regardless of the methodology used, early behavioural assessments of the child reach their limits when it comes to developing a reliable and accurate diagnosis. This is due to the lack of stability in symptom expression during the first months of development, and the difficulty of distinguishing ASD from other severe developmental disorders, such as

language disorders (dysphasia) or intellectual disabilities, before 24 months of age. During this period, infants do not have the developmental tools to fully express all the signs of ASD (Saint-Georges et al., 2013).

In the absence of biological markers, early screening cannot provide a precise and positive diagnosis of ASD, but can only identify children at risk of later developmental issues, regardless of the quality of the tools used. Diagnostic errors, whether due to rushed assessments or difficulties in distinguishing between similar syndromes, can have serious consequences. These include the psychological suffering of the family, social stigma related to the disorder, and the proposal of inappropriate therapeutic and rehabilitative approaches. Such errors can even affect the child's future, as critical decisions made early on may significantly limit their chances of later social, educational, and professional integration. This situation is exacerbated by the presence of unqualified professionals and the use of inappropriate assessment and diagnostic tools.

3. Autism Spectrum Disorder and the Challenge of Epidemiological Characteristics:

The diagnostic criteria for autism have evolved and varied significantly over time, starting from the standards introduced by Kanner, moving through those adopted by Rutter, and continuing with the various global classifications, including the eleventh edition of the World Health Organization's International Classification and the fifth edition of the American Psychiatric Association's Diagnostic and Statistical Manual (DSM-5). The early criteria primarily reflected more severe manifestations, particularly those involving significant delays in language and cognitive skills.

In the 1980s, less severe forms of ASD, which were not associated with intellectual disability, were integrated into the spectrum, such as pervasive developmental disorders not otherwise specified (PDD-NOS). Asperger's syndrome also emerged in the 1990s. Some subtypes of autism spectrum disorders, such as residual autism, were identified in the third edition of the DSM (DSM-3) but were later removed in subsequent editions.

The variation in epidemiological study results can also be attributed to differences in the sources of data used for detection. Some studies relied solely on specialized databases, others on special education records, and some on national registries. These sources share a common limitation: they focus on previously diagnosed populations rather than the general population, thus excluding individuals with autism who have not been in contact with these services, leading to an underestimation of prevalence (Fombonne et al., 2019).

It is also important to note that after case detection, a subsequent stage involves diagnosis confirmation, drawing on multiple data sources (parents, teachers, doctors, medical files, and educational records) as well as assessments of the person suspected of having autism. The challenge is that this comprehensive approach is not available for all studies. Additionally, the lack of adequately trained professionals, limited access to diagnostic resources, and poorly adapted evaluation tools significantly contribute to the misestimation of epidemiological characteristics.

In line with the variability of epidemiological data, autism prevalence rates have been steadily increasing over the years. This rise became particularly noticeable after Asperger's syndrome was included within the autism spectrum in the 1990s. Several explanations have been proposed to account for this significant increase. Some studies linked the rise to the consequences of the MMR (measles, mumps, and rubella) vaccine, suggesting that exposure to it may have led to a type of autism characterized by gastrointestinal disturbances and developmental regression (Wakefield et al., 1998). Similar claims were made regarding vaccines containing arsenic, known for its toxicity and impact on the central nervous system. However, a large-scale Danish study conducted by Hviid and colleagues (2019) followed over 650,000 Danish children born to Danish mothers between 1999 and 2010 until 2013. The study considered many factors, including family history of autism, parental age, smoking during pregnancy, potential premature birth, Apgar scores (a measure of newborn vitality), birth weight, and head circumference. The study confirmed that the MMR vaccine does not increase the risk of autism, nor does it cause autism in children with risk factors or lead

to autism clusters after vaccination (Hviid et al., 2019).

In another review, Fombonne (2019) highlighted methodological and contextual factors that could explain the increase in autism prevalence. He pointed out that the rise could be attributed to changes in diagnostic algorithms, the continuous expansion of criteria and diagnostic tools, earlier detection, and improved identification of cases. Additionally, the reclassification of children who were previously diagnosed with intellectual disabilities or language disorders as having ASD has contributed to the rise in prevalence. For example, individuals with Asperger's syndrome were previously diagnosed with obsessive-compulsive disorder (OCD), school phobia, or social anxiety before being recognized as having an autism spectrum disorder. In some countries, obtaining an autism diagnosis instead of an intellectual disability diagnosis makes it easier to access specialized care programs (Cohen et al., 2012).

Another issue in the epidemiological characteristics is the gender variable, as autism has long been considered more prevalent in males. According to the DSM-5, males are diagnosed with ASD at a rate four times higher than females (APA, 2013). However, recent research indicates that ASD in females may be significantly underdiagnosed, particularly when they do not have co-occurring intellectual disabilities or language impairments. Girls who do not exhibit these deficits may not meet the diagnostic criteria for autism. Many researchers link this gender bias to the way autism studies have historically been conducted. From the time Kanner and Asperger described the symptoms, the focus was primarily on males. For instance, among the four individuals studied by Asperger, all were boys. In Kanner's 11-case study, only three were girls. Since then, most autism research has focused on males, and the behaviours described in the literature are derived mainly from observations of boys or men with autism. In recent years, however, studies addressing the specific female autism phenotype have emerged, gathering more evidence to support this concept.

Other studies have sought to explain these gender differences by pointing to potential protective factors—genetic, hormonal, or related to brain plasticity—or even environmental factors related to social upbringing and gender expectations. Females with autism often develop coping and masking strategies that may help them integrate socially, even though these strategies are often exhausting and have serious long-term consequences. This may reduce their chances of early diagnosis and limit their access to appropriate interventions and services. Similarly, their restricted interests and stereotyped behaviours may align more closely with societal expectations (e.g., interests in animals, dance, or fantasy worlds). Additionally, girls may perform better on executive function tasks (such as cognitive flexibility, information processing speed, and self-generated responses), especially when these tasks are assessed through standardized tests. This can mask some difficulties that might otherwise hinder everyday functioning and affect their independence.

Among the theories that favour the diagnosis of autism in males while downplaying its prevalence in females is the "extreme male brain" theory proposed by Baron-Cohen (2002, 2009). This theory posits that ASD represents an extreme version of the male brain, which excels at systemizing but struggles with empathy. Research suggests that females with autism are better at recognizing facial emotional expressions than males with autism. While this theory is supported by numerous clinical and experimental arguments from neuroscience, it may also have contributed to an under appreciation of empathic abilities in males and organizational abilities in females, despite both being present in both genders.

4. Autism Spectrum Disorder and the Issue of Causality:

Current studies largely emphasize genetic and biochemical factors that influence the central nervous system at an early stage, leading to the symptoms of autism spectrum disorder (ASD) and related conditions. Despite significant efforts and advancements in understanding ASD across various psychological, cognitive, and neurobiological fields, no clear and precise causality has been established that can unravel the mystery of autism symptoms and the diverse clinical features associated with it.

For forty years, psychoanalytic theory dominated, focusing primarily on early

interactions between the infant and the mother, maternal bonding, and the responsibility of parents—especially the mother—for the development of autism in their children. However, these concepts were largely undermined in the 1990s.

Today, biological hypotheses, particularly genetic and biochemical ones have gained widespread support among professionals (Jamain et al., 2003). In general, most genetic studies suggest a hereditary predisposition to autism. However, no single primary gene has been identified. The significant variation in autism disorders on the one hand, the large number of discovered mutations on the other, and the heterogeneity of the results have made it difficult for researchers to pinpoint specific genes. Instead, these studies indicate substantial genetic variation within the syndrome. The complexity increases when it becomes apparent that 10-20% of genetic mutations in those affected are not inherited but arise spontaneously (Zerouali, 2021).

Similarly, despite the extensive research on biochemical hypotheses, the results remain inconclusive and subject to debate due to several methodological challenges. The heterogeneity of the findings could be attributed to multiple influences such as age, gender, and diet. Additionally, a lack of strict methodological controls and small sample sizes may have led to findings that could not be replicated later, or conversely, subtle differences between individuals with autism and control groups may have gone unnoticed (Zerouali, 2021).

Given the diversity of cases and clinical presentations, and because autism is defined primarily through behavioural criteria, generalizations are difficult to make. The pathogenic causes could be manifold: prenatal or postnatal incidents, hereditary predispositions, metabolic issues—all of these could contribute to the development of autism symptoms. Although less than a quarter of autism cases are currently linked to specific conditions, the causes of the remaining three-quarters remain unknown (Jamain et al., 2003).

Approximately 15% of autism cases are associated with genetic conditions such as Fragile X syndrome, Tuberous Sclerosis, Rett syndrome, or Angelman syndrome. Despite this variety, debates continue between proponents of different theories of autism's origins, such as genetics, toxins, environmental pollution, and vaccines.

Currently, it is difficult to determine whether there is a cause-and-effect relationship between the biological disorders faced by children with autism and their cognitive and clinical symptoms, and if so, in which direction this relationship operates. Therefore, while the existence of biological factors cannot be denied, reducing the causes of autism to purely biological models that overlook environmental influences is not advisable. Ultimately, autism in childhood can be seen as a pathological psychological structure that may develop in response to a wide range of initial factors, both organic and psychological.

Taking this multifactorial aspect into account, a multidisciplinary approach to investigating the causes of autism could prove highly beneficial, which, in turn, could inform the development of appropriate and multidimensional therapeutic approaches (Zerouali, 2021).

5. Autism Spectrum Disorder and the Therapeutic Challenges:

In response to the various theories explaining autism, numerous therapeutic, rehabilitative programs, and dietary interventions have emerged. These programs reflect a mix of good intentions to help children with autism and their families find the best ways to cope with the disorder, alongside exploitative attempts by some to profit from expensive and often ineffective therapies or dietary regimens. Recent research has shown that many of these treatments, including alternative therapies, are ineffective, leaving families confused and struggling without finding effective, adaptable solutions.

For a long time, psychoanalytic treatments dominated the field, but they were the subject of significant controversy, leading to resistance from many parent associations and conflicts between professionals who adhered to this therapeutic model. Many specialists view psychodynamic approaches, which focus on understanding the meaning behind symptoms, as contributing to a better understanding of the mental lives of individuals with autism, and thus helping to tailor therapeutic approaches more

effectively. This approach, grounded in psychoanalysis, involves a variety of professionals working to stimulate exchanges through different interventions (therapeutic, educational, sports, and social interactions) (Dellion, 2011). However, psychoanalytic therapy has been largely abandoned under pressure from parent associations, who opposed its emphasis on maternal responsibility and its strategy of separating the child from the family unit.

Since the 1990s, the expansion of diagnostic criteria for autism has fuelled research in genetics, neuroimaging, cognitive science, neuroscience, and more. Simultaneously, a large market has developed, including medications, behavioural methods, special diets, and genetic testing. For many, autism has become a significant public health issue, garnering widespread media attention. The variety of therapeutic and rehabilitative approaches reflects the growing demand, but there remains a significant gap in the research confirming the effectiveness of these interventions. When studies do exist, they often suffer from methodological biases related to sample selection, size, and protocol variables.

Currently, behavioural programs such as TEACCH (Treatment and Education of Autistic and Related Communication Handicapped Children) and Applied Behaviour Analysis (ABA) dominate. ABA is particularly intensive, involving 40 hours of therapy per week, and aims to maximize the child's independence using operant conditioning and repeated trials (repeating activities until the desired results are achieved). Positive reinforcements (such as smiles, praise, or treats) are used to encourage appropriate behaviours. These programs focus on the emerging skills of children and involve collaboration between parents and professionals, with parents receiving training to implement these strategies at home. Despite extensive media coverage and widespread popularity, ABA has been heavily criticized for its high cost (requiring one specialist per child, under the supervision of a psychologist) (Chamak, 2020). Moreover, additional studies are needed to confirm its effectiveness and address ethical concerns regarding certain coercive techniques used in the program. Even the program's founders have raised concerns about the methodology and general criteria applied, noting that the progress seen in children might result more from compliance and learning rather than natural cognitive development (Schopler).

The same applies to other therapeutic approaches, such as the Son-Rise Program, the Makaton method, play therapy, sensory-motor therapies, and alternative treatments involving various dietary interventions. While these methods have garnered significant media attention, the results are still the subject of debate and varied opinions. For example, the Son-Rise Program, developed by Barry Neil Kaufman and his wife Samahria Lyte Kaufman for their son with autism, gained widespread attention when their son reportedly recovered fully from autism. The program focuses on increasing spontaneous social orientation, communication, and social interaction skills. However, a study by Williams and Wishaft (2003) found more shortcomings than benefits with this approach. Additionally, France's (Haute Autorité de Santé) (HAS, 2012) recommended against its use, citing a lack of evidence regarding its effectiveness and a weak theoretical foundation.

Similarly, the (National Institute for Health and Care Excellence) (NICE, 2013) in the UK has emphasized that there is no cure for autism. Instead, there are interventions that may address some symptoms, behaviours, and issues related to the disorder (Chamak, 2017).

Furthermore, many guidelines stress the importance of improving the quality of life for children with autism and their families by providing accessible health and social services and encouraging play-based interventions to support joint attention and communication skills. Additionally, they advocate for addressing related physical problems, such as gastrointestinal disorders and psychological issues like anxiety, depression, and hyperactivity.

6. Conclusion:

Despite the various challenges mentioned, there is a consensus among many researchers and practitioners on certain common foundations, particularly the importance of early diagnosis and intervention, as well as the significance of the relationships between parents and professionals. To address the issues related to diagnosis, treatment, and rehabilitation, a multidisciplinary approach is essential,

integrating therapeutic, rehabilitative, and educational aspects. Additionally, comprehensive and ongoing training for specialists is critical and must be considered in any strategy that aims to be effective and credible, whether in detection, diagnosis, or treatment.

Moreover, adhering to ethical guidelines is crucial, as it obliges professionals to acknowledge the limits of their knowledge, methodology, and expertise, while respecting the rights of the child and responding to their suffering without compromising their dignity and humanity. Raising awareness programs for parents is also key. These programs not only support professionals in accurate and effective diagnosis by helping parents develop the ability to recognize early signs of autism, but also assist parents in understanding the problem and its associated difficulties. In addition, parents can play an active role in treatment and integration procedures.

Currently, a wealth of information supports making collaboration between parents and professionals both possible and essential, which may significantly reduce the psychological burden on families. It is self-evident that families should have a say in the support systems provided for their children. This should be done within a framework of mutual respect and trust, avoiding, as much as possible, the dynamics of dominance and authority that professionals may impose on the parents of children with autism under the pretext of exclusive knowledge and treatment capabilities.

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