

# American Journal of Psychology and Brain Studies

<https://urfpublishers.com/journal/american-psychology>

Vol: 3 & Iss: 1

## Autism Spectrum Disorder in Children: Current Perspectives on Etiology, Diagnosis and Early Intervention

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**Citation:** Manseur N. Autism Spectrum Disorder in Children: Current Perspectives on Etiology, Diagnosis and Early Intervention. *Am J Psychol & Brain Stud*, 2026;3(1):199-203.

**Received:** 09 March, 2026; **Accepted:** 16 March, 2026; **Published:** 18 March, 2026

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### ABSTRACT

Autism Spectrum Disorder (ASD) is a complex neurodevelopmental condition characterized by persistent impairments in social communication and the presence of restricted, repetitive behaviors and interests. Over the past several decades, the global recognition and diagnosis of ASD have increased substantially, making it one of the most significant developmental disorders affecting children worldwide. The growing prevalence of autism has generated extensive scientific interest in understanding its etiology, early detection, and intervention strategies.

This review article synthesizes current scientific knowledge on Autism Spectrum Disorder in children, focusing on epidemiology, etiological factors, clinical manifestations, diagnostic approaches, and intervention strategies. Current evidence suggests that ASD arises from a multifactorial interaction of genetic, neurobiological, and environmental influences affecting early brain development. Advances in neuroimaging and genetics have contributed significantly to understanding the neurodevelopmental mechanisms underlying the disorder.

Early identification of ASD remains critical for improving developmental outcomes. Research consistently demonstrates that early behavioral and educational interventions significantly enhance language development, social functioning, and adaptive skills. Multidisciplinary approaches involving healthcare professionals, educators, and families are essential for optimizing therapeutic outcomes.

Despite considerable progress in ASD research, significant challenges remain in understanding the heterogeneity of the disorder and developing personalized treatment strategies. Continued research is necessary to improve early detection methods, clarify underlying biological mechanisms, and expand effective intervention programs for children with ASD.

**Keywords:** Autism Spectrum Disorder; Child Development; Neurodevelopmental Disorders; Early Intervention; Behavioral Therapy; Developmental Psychology

## 1. Introduction

Autism Spectrum Disorder (ASD) is widely recognized as one of the most complex neurodevelopmental conditions affecting children. Characterized primarily by persistent difficulties in social communication and interaction, as well as restricted and repetitive patterns of behavior, ASD presents a wide range of clinical manifestations that vary significantly in severity and functional impact. According to the Diagnostic and Statistical Manual of Mental Disorders (DSM-5-TR), ASD encompasses a spectrum of developmental impairments that typically emerge during early childhood and influence cognitive, social, and behavioral functioning throughout the lifespan<sup>1</sup>.

Over the past several decades, the prevalence of ASD has increased substantially across many regions of the world. While part of this increase can be attributed to improved diagnostic awareness and expanded diagnostic criteria, research suggests that the growing recognition of autism also reflects greater access to developmental screening and heightened awareness among healthcare professionals and parents. Recent global estimates indicate that approximately one in one hundred children is diagnosed with ASD, although prevalence rates may vary across different populations and diagnostic methodologies<sup>2</sup>.

Autism is not a single uniform disorder but rather a heterogeneous condition characterized by considerable variability in symptom severity, cognitive abilities, and adaptive functioning. Some children with ASD experience significant intellectual disability and limited verbal communication, whereas others possess average or even superior intellectual abilities but face challenges in social interaction and pragmatic communication. This variability has led researchers to conceptualize autism as a spectrum disorder, acknowledging the diverse range of developmental profiles associated with the condition.

The early years of life represent a critical period for brain development, during which neural circuits responsible for language, social cognition, and emotional regulation undergo rapid maturation. Disruptions in these developmental processes are believed to contribute to the emergence of ASD symptoms. Consequently, early identification and intervention have become central priorities in autism research and clinical practice.

The importance of early detection cannot be overstated. Numerous studies have demonstrated that early therapeutic interventions can significantly improve language acquisition, social engagement, and adaptive behavior in children with ASD. Behavioral therapies, speech and language interventions, occupational therapy, and educational support programs all play essential roles in helping children reach their developmental potential.

This article provides a comprehensive review of current scientific knowledge regarding Autism Spectrum Disorder in children. The discussion focuses on the epidemiology of autism, its potential etiological factors, clinical manifestations, diagnostic procedures, and evidence-based intervention strategies. By synthesizing existing research, this review aims to provide a clearer understanding of ASD and highlight the importance of early intervention and multidisciplinary care.

## 2. Epidemiology of Autism Spectrum Disorder

Understanding the epidemiology of autism spectrum disorder is essential for appreciating its impact on global child

health. Epidemiology examines the distribution, prevalence, and determinants of diseases within populations, offering valuable insights into patterns of diagnosis and potential risk factors.

Over the past three decades, epidemiological studies have documented a steady increase in the reported prevalence of ASD. Early estimates from the 1980s suggested that autism affected approximately four to five children per 10,000. In contrast, contemporary studies report significantly higher prevalence rates. Surveillance data from several developed countries now estimate that approximately one in thirty-six children may meet diagnostic criteria for ASD.

Multiple factors have contributed to this apparent rise in prevalence. One major factor is the broadening of diagnostic criteria. Earlier diagnostic frameworks focused primarily on classic autism characterized by severe impairments in communication and social functioning. However, modern diagnostic systems recognize a broader spectrum of related developmental conditions, including milder forms of autism that might previously have gone unrecognized.

Increased public awareness has also played a critical role. Parents, educators, and healthcare providers are now more familiar with early warning signs of developmental delays. Routine developmental screening during pediatric visits has further improved the detection of autism symptoms in young children.

Gender differences in ASD prevalence represent another consistent epidemiological finding. Numerous studies indicate that boys are diagnosed with autism more frequently than girls, with male-to-female ratios typically ranging from 3:1 to 4:1. The reasons for this disparity remain a subject of ongoing research. Some scientists suggest that biological factors may increase vulnerability among males, while others propose that autism in girls may be underdiagnosed due to differences in symptom presentation.

Socioeconomic and cultural factors may also influence diagnostic patterns. Access to healthcare services, educational resources, and early intervention programs can vary widely across countries and communities. In regions with limited diagnostic infrastructure, many children with developmental disorders may remain undiagnosed or misdiagnosed.

Despite these variations, the global burden of ASD is increasingly recognized as a significant public health concern. Children with autism often require long-term educational and therapeutic support, and families frequently face emotional, financial, and social challenges related to caregiving responsibilities.

For these reasons, many governments and international health organizations have prioritized early detection and intervention strategies aimed at improving outcomes for children with ASD. Continued epidemiological research is essential for identifying trends, informing policy decisions, and guiding the allocation of healthcare resources.

## Etiology and Neurodevelopmental Mechanisms

The etiology of autism spectrum disorder remains one of the most complex and actively investigated topics in developmental neuroscience. Rather than resulting from a single cause, ASD is widely understood to arise from the interaction of multiple

genetic, neurobiological, and environmental factors that influence early brain development.

Genetic influences represent one of the most significant contributors to ASD risk. Twin studies have provided compelling evidence for the heritability of autism. Research comparing identical and fraternal twins suggests that genetic factors account for a substantial proportion of ASD susceptibility. Advances in molecular genetics have identified numerous genes associated with neural development, synaptic function, and neuronal connectivity that may contribute to autism risk.

However, no single gene can fully explain the disorder. Instead, ASD appears to involve complex interactions among multiple genetic variants. Some of these variants influence the formation of synapses—the connections between neurons—while others affect the regulation of gene expression during early brain development.

Neuroimaging studies have provided additional insights into the neurological basis of autism. Brain imaging research has revealed differences in the structure and connectivity of several brain regions involved in social cognition, language processing, and emotional regulation. For example, atypical patterns of connectivity have been observed in the prefrontal cortex, amygdala, and temporal lobes, areas that play key roles in social perception and communication.

Environmental influences may also contribute to ASD risk, particularly during prenatal development. Factors such as maternal infections during pregnancy, exposure to certain environmental toxins, and complications during childbirth have been investigated as potential contributors. While these factors alone do not cause autism, they may interact with genetic susceptibility to influence neurodevelopmental outcomes.

Importantly, extensive scientific research has found no credible evidence linking vaccines to autism. This misconception originated from a now-discredited study and has been thoroughly refuted by large-scale epidemiological investigations involving millions of children.

The emerging consensus among researchers is that ASD results from disruptions in early brain development affecting neural circuits responsible for social communication and cognitive flexibility. These disruptions may occur during prenatal development or early infancy, long before behavioral symptoms become visible.

Understanding these biological mechanisms is essential for developing more effective diagnostic tools and therapeutic interventions.

### **Clinical Characteristics of Autism Spectrum Disorder in Children**

Autism Spectrum Disorder manifests through a wide range of behavioral and developmental characteristics that typically emerge during early childhood. Although the specific presentation of symptoms varies considerably among individuals, clinicians generally recognize two primary diagnostic domains: deficits in social communication and interaction, and restricted or repetitive patterns of behavior, interests, or activities.

One of the earliest observable features of ASD is difficulty in social engagement. Infants who later receive an autism diagnosis may display reduced eye contact, limited response to their name,

and diminished interest in social interaction with caregivers. These early signs often become more pronounced during the second year of life, when developmental milestones related to language and social communication typically accelerate.

Children with ASD frequently exhibit impairments in nonverbal communication. Gestures such as pointing, waving, or nodding may be absent or used atypically. Facial expressions and body language may also be less responsive or poorly synchronized with social contexts. These differences can make reciprocal interaction with peers and adults more challenging.

Language development in children with autism can vary widely. Some children experience significant delays in speech acquisition, while others develop extensive vocabularies but struggle with pragmatic language skills such as conversational turn-taking, understanding figurative expressions, or interpreting tone and context. Echolalia, the repetition of words or phrases spoken by others, is a common feature observed in some children with ASD.

Restricted and repetitive behaviors represent another core characteristic of the disorder. These behaviors may include repetitive motor movements such as hand-flapping, rocking, or spinning objects. Children with ASD often demonstrate a strong preference for routines and may become distressed when familiar patterns or environments change unexpectedly.

Many individuals with autism also exhibit restricted interests that are unusually intense or highly focused. A child might become deeply absorbed in specific topics such as numbers, maps, mechanical systems, or particular objects. While these interests can sometimes support learning and cognitive development, they may also limit engagement with broader social experiences.

Sensory processing differences are also frequently reported among children with ASD. Some children may display hypersensitivity to sounds, lights, textures, or smells, while others may seek sensory stimulation through repetitive behaviors. These sensory experiences can significantly influence behavior and daily functioning.

Comorbid conditions are common in ASD. Many children with autism also experience additional developmental or psychiatric conditions, including attention-deficit/hyperactivity disorder (ADHD), anxiety disorders, sleep disturbances, epilepsy, or intellectual disability. These co-occurring conditions can complicate diagnosis and treatment, emphasizing the importance of comprehensive clinical assessment.

The variability in symptom presentation highlights the importance of individualized evaluation and intervention strategies. Recognizing the diversity within the autism spectrum allows clinicians and educators to design supportive environments that address each child's unique developmental profile.

### **Diagnostic Approaches and Screening**

Accurate and timely diagnosis of autism spectrum disorder is essential for initiating early intervention and providing appropriate support for affected children and their families. Modern diagnostic practices rely primarily on behavioral assessments guided by standardized diagnostic criteria and structured evaluation tools.

The Diagnostic and Statistical Manual of Mental Disorders

(DSM-5-TR) defines ASD based on two primary domains: persistent deficits in social communication and social interaction, and restricted or repetitive patterns of behavior. Symptoms must be present in early developmental stages and must significantly impair social, occupational, or other important areas of functioning.

Early screening plays a critical role in identifying children who may require further evaluation. Pediatric developmental surveillance typically occurs during routine health visits in early childhood. Healthcare providers monitor developmental milestones such as language acquisition, social responsiveness, and motor development.

One widely used screening instrument is the Modified Checklist for Autism in Toddlers (M-CHAT), which is designed to identify early signs of autism in children between 16 and 30 months of age. The screening questionnaire is completed by parents and focuses on behaviors related to joint attention, social engagement, and communication.

When screening results suggest developmental concerns, a comprehensive diagnostic evaluation is recommended. Such evaluations are usually conducted by multidisciplinary teams that may include pediatricians, child psychiatrists, psychologists, speech-language pathologists, and occupational therapists.

Among the most widely recognized diagnostic tools are the Autism Diagnostic Observation Schedule (ADOS-2) and the Autism Diagnostic Interview-Revised (ADI-R). These instruments provide structured observations and detailed caregiver interviews that assess communication patterns, social behavior, and repetitive activities.

Advances in neuroscience have also introduced new research approaches aimed at identifying biological markers associated with autism. Neuroimaging studies, electroencephalography (EEG), and genetic analyses are currently being explored as potential complementary tools for understanding the neurobiological mechanisms underlying ASD. However, these techniques remain primarily within the research domain and are not yet widely used in routine clinical diagnosis.

Early diagnosis allows children with ASD to access intervention services during critical periods of brain development. Research consistently demonstrates that children who begin therapeutic programs at younger ages tend to experience greater improvements in communication, adaptive behavior, and social functioning.

For this reason, international health organizations strongly recommend universal developmental screening during early childhood. Increased awareness among parents, educators, and healthcare providers is essential for ensuring that children with autism receive timely evaluation and support.

### **Early Intervention and Therapeutic Approaches**

Early intervention represents one of the most significant factors influencing developmental outcomes for children with autism spectrum disorder. Because the human brain undergoes rapid development during the first years of life, therapeutic interventions initiated during early childhood can significantly enhance language acquisition, social engagement, and adaptive functioning.

Behavioral therapy is among the most widely studied and implemented approaches for supporting children with ASD. One of the most well-known models is Applied Behavior Analysis (ABA), which focuses on reinforcing positive behaviors and teaching new skills through structured learning experiences. ABA-based interventions aim to improve communication, social interaction, and daily living skills by breaking complex behaviors into smaller, teachable components.

Speech and language therapy plays a crucial role in addressing communication challenges associated with autism. Speech-language pathologists work with children to develop expressive and receptive language skills, improve articulation, and enhance pragmatic communication abilities. For children with limited verbal communication, alternative communication systems such as picture exchange communication systems (PECS) or speech-generating devices may be introduced.

Occupational therapy is another essential component of multidisciplinary intervention programs. Occupational therapists help children develop fine motor skills, sensory integration abilities, and functional independence in daily activities such as dressing, eating, and personal hygiene.

Educational interventions are also critical for supporting children with ASD. Specialized educational programs often incorporate structured teaching methods, visual supports, and individualized learning plans tailored to the child's developmental level and learning style. Collaborative partnerships between educators, therapists, and families are essential for ensuring consistency across home and school environments.

Family involvement is a cornerstone of effective intervention. Parents and caregivers play a vital role in reinforcing therapeutic strategies and creating supportive learning environments. Parent-mediated interventions have shown promising results in improving communication and social interaction skills.

While no single intervention approach is universally effective for all children with ASD, research consistently supports the value of comprehensive, individualized programs that integrate behavioral, educational, and developmental strategies.

### **Psychosocial Impact on Families**

Autism Spectrum Disorder affects not only the individual child but also the broader family system. Parents and caregivers often face emotional, financial, and social challenges associated with raising a child with developmental differences.

The process of obtaining an autism diagnosis can be emotionally complex for families. Many parents initially experience uncertainty, confusion, or concern as they navigate the diagnostic process and seek information about the condition. Access to accurate information and supportive healthcare professionals can help families better understand the developmental needs of their child.

Caregiving responsibilities may be demanding, particularly when children require intensive therapeutic interventions or specialized educational support. Families may need to coordinate multiple therapy appointments, educational services, and medical consultations, which can create logistical and financial pressures.

Despite these challenges, many families also report positive experiences related to resilience, personal growth, and strengthened family relationships. Support networks, community resources, and parent support groups can provide valuable emotional and practical assistance.

Recognizing the importance of family well-being, many intervention programs now incorporate family-centered approaches that provide guidance, counseling, and education for caregivers. Supporting families is essential for promoting positive outcomes for children with ASD.

### **Future Directions in Autism Research**

Although significant progress has been made in understanding autism spectrum disorder, many questions remain unanswered. Future research will likely focus on several key areas.

Advances in genetics and neuroscience are expected to deepen our understanding of the biological mechanisms underlying ASD. Identifying specific genetic pathways and neural networks involved in autism may eventually support the development of targeted therapeutic interventions.

Another important research direction involves improving early detection methods. Scientists are exploring the use of machine learning algorithms, digital behavioral analysis, and neurophysiological markers to identify subtle early indicators of autism before behavioral symptoms become fully apparent.

Personalized intervention strategies also represent a promising area of research. Because ASD is highly heterogeneous, individualized treatment plans that consider each child's unique developmental profile may enhance therapeutic outcomes.

In addition, increasing attention is being given to the long-term experiences of individuals with autism, including adolescence and adulthood. Supporting individuals across the lifespan requires expanded educational opportunities, vocational training, and social inclusion initiatives.

Continued interdisciplinary collaboration among researchers, clinicians, educators, and policymakers will be essential for advancing knowledge and improving support systems for individuals with autism and their families<sup>3-5</sup>.

### **Conclusion**

Autism Spectrum Disorder represents a complex and multifaceted neurodevelopmental condition that affects millions of children worldwide. Characterized by differences in social communication, behavior, and sensory processing, ASD presents significant challenges but also highlights the diversity of human development.

Over the past several decades, substantial progress has been made in understanding the biological, psychological, and social aspects of autism. Increased awareness, improved diagnostic tools, and expanded early intervention programs have contributed to better developmental outcomes for many children.

Early identification remains one of the most critical factors in supporting children with ASD. When intervention begins during the early stages of development, children often demonstrate significant improvements in language, social interaction, and adaptive functioning.

A multidisciplinary approach involving healthcare professionals, educators, therapists, and families is essential for addressing the diverse needs of children with autism. By combining scientific research with compassionate clinical practice, societies can continue to improve the quality of life for individuals with ASD and their families.

Future research will play a vital role in advancing our understanding of autism and developing innovative strategies for early detection, personalized intervention, and lifelong support.

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