



## Autism Spectrum Disorder: Psychological Assessment, Diagnosis, and Intervention-An Updated Review

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### Abstract:

**Background:** Autism Spectrum Disorder (ASD) is a lifelong neurodevelopmental condition defined by impairments in social communication and restricted, repetitive patterns of behavior. Its prevalence has increased globally due to improved diagnostic awareness, broadened clinical criteria, and enhanced surveillance systems. Despite extensive research, ASD remains a highly heterogeneous condition with multifactorial etiology and variable clinical outcomes.

**Aim:** This paper aims to provide a comprehensive synthesis of ASD covering diagnostic criteria, epidemiology, etiology, pathophysiology, clinical evaluation, management approaches, prognosis, and associated complications.

**Methods:** A narrative review approach was adopted using evidence-based literature, clinical guidelines, and DSM-5-TR diagnostic criteria. The review integrates findings from genetic, neurobiological, environmental, and clinical research to present a holistic understanding of ASD. **Results:** ASD is associated with strong genetic contributions, including synaptic and transcriptional gene pathways, alongside environmental risk factors such as prenatal exposures and perinatal complications. Neurobiological findings suggest altered brain connectivity, atypical synaptogenesis, and abnormal developmental trajectories. Diagnosis is clinical, supported by standardized tools and behavioral assessment. Management requires individualized, multidisciplinary interventions incorporating behavioral therapies, educational support, and pharmacological treatment for comorbid conditions. Prognosis varies widely and is influenced by early intervention, cognitive ability, and family support. Complications frequently include psychiatric disorders, epilepsy, sleep disturbances, and intellectual disability. **Conclusion:** ASD is a multifactorial condition requiring early identification and lifelong, integrated management strategies. A shift toward neurodiversity-informed care emphasizes both support needs and individual strengths, improving functional outcomes and quality of life.

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## Introduction

Autism Spectrum Disorder (ASD) is recognized as a complex and heterogeneous neurodevelopmental condition characterized by persistent challenges in social communication and interaction, accompanied by restricted, repetitive patterns of behavior, interests, and activities. The condition encompasses a broad spectrum of presentations, reflecting substantial variability in symptom severity, cognitive functioning, language abilities, adaptive skills, and associated comorbidities. This diversity in clinical manifestation underscores the need for a comprehensive diagnostic framework capable of capturing the multidimensional nature of the disorder. The Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition, Text Revision (DSM-5-TR) provides such a framework by establishing standardized diagnostic criteria that emphasize both social communication impairments and patterns of restricted or repetitive behavior as the defining features of ASD [1]. A fundamental characteristic of ASD involves persistent deficits in social communication and social interaction that occur across various contexts and developmental stages. These impairments extend beyond isolated communication difficulties and encompass a broad range of social challenges that affect interpersonal functioning. Individuals with ASD frequently experience limitations in social-emotional reciprocity, which may manifest as difficulties initiating conversations, maintaining reciprocal exchanges, sharing interests and emotions, or appropriately responding to social interactions. The severity of these difficulties varies considerably among affected individuals. Some may demonstrate subtle impairments in conversational reciprocity and emotional sharing, whereas others may exhibit profound challenges that significantly limit their ability to engage in meaningful social relationships. These deficits often interfere with the development of effective interpersonal communication skills and may contribute to social isolation and reduced participation in community and educational environments [1].

In addition to challenges related to social reciprocity, individuals with ASD frequently display impairments in nonverbal communication behaviors that are essential for successful social interaction. Nonverbal communication constitutes a critical component of human social functioning, facilitating the expression and interpretation of emotions, intentions, and interpersonal cues. Individuals with ASD may experience difficulties integrating verbal and nonverbal communication effectively, resulting in atypical patterns of eye contact, body language, facial expression, and gesture use. These challenges may range from subtle abnormalities in social signaling to profound impairments characterized by limited or absent use of nonverbal communication. Such difficulties can hinder social understanding and contribute to misunderstandings during interpersonal interactions, further complicating the establishment and maintenance of social relationships [1]. Another defining aspect of ASD involves persistent difficulties in developing, maintaining, and understanding interpersonal relationships. These impairments often become evident during childhood and may continue throughout adolescence and adulthood. Individuals with ASD may encounter challenges adapting their behavior to different social contexts, participating in imaginative or cooperative play, establishing friendships, and understanding the perspectives and intentions of others. In some cases, reduced interest in peer relationships may be observed, while in others, a desire for social engagement exists but is hindered by deficits in social cognition and communication. The cumulative effect of these challenges can significantly impact educational achievement, occupational functioning, and overall quality of life, emphasizing the importance of early identification and intervention [1].

Alongside social communication impairments, ASD is characterized by restricted and repetitive patterns of behavior, interests, and activities. These behavioral manifestations constitute a core diagnostic domain and contribute substantially to the functional profile of individuals with autism. Repetitive motor movements, stereotyped behaviors, and repetitive use of objects are commonly observed. Examples include hand flapping, rocking, repetitive manipulation of objects, echolalia, and the use of idiosyncratic phrases. Such behaviors may serve various functions, including self-regulation, sensory stimulation, anxiety reduction, or communication. While these behaviors can sometimes be adaptive, they may also interfere with daily functioning and social participation when they become excessive or inflexible [1]. A pronounced preference for sameness and predictability is another hallmark feature of ASD. Many individuals demonstrate rigid adherence to routines, rituals, and established patterns of behavior. Even minor alterations in daily routines or environmental circumstances may provoke significant distress, anxiety, or behavioral disruption. Difficulties with transitions, inflexible thinking patterns, ritualized behaviors, and persistent reliance on familiar routines are commonly reported. This need for consistency reflects underlying differences in cognitive processing and adaptation to change. Consequently, unexpected events or disruptions in routine may present substantial challenges, affecting both individual functioning and family dynamics [1]. Restricted and highly focused interests also represent a central component of the autism phenotype. Individuals with ASD often develop intense interests in specific topics, objects, or activities that exceed typical levels of engagement in both depth and duration. These interests may involve highly specialized subjects and can become a primary source of motivation, enjoyment, and expertise. While such interests can facilitate learning and skill development, they may also limit engagement in

broader activities and social experiences when they dominate attention and daily functioning. The intensity and specificity of these interests frequently distinguish them from typical hobbies or preferences observed in the general population [1].

Sensory processing differences constitute another significant dimension of ASD. Individuals may demonstrate either heightened sensitivity or reduced responsiveness to sensory stimuli, including auditory, visual, tactile, olfactory, gustatory, and proprioceptive inputs. Sensory experiences that appear ordinary to others may be perceived as overwhelming, distressing, or uncomfortable by autistic individuals. Conversely, some individuals may exhibit diminished awareness of sensory stimuli such as pain or temperature. Unusual sensory interests, including fascination with lights, movement, textures, sounds, or smells, are also frequently reported. These sensory processing differences can substantially influence behavior, emotional regulation, learning experiences, and participation in daily activities, highlighting the importance of sensory-informed approaches in clinical and educational settings [1]. The DSM-5-TR further emphasizes that ASD symptoms must emerge during the early developmental period, although their full manifestation may not become apparent until social demands exceed an individual's capacities. During early childhood, developmental differences may be subtle or masked by supportive environments. As social expectations increase with age, difficulties in communication, social interaction, and behavioral flexibility often become more evident. Some individuals develop compensatory strategies that partially conceal symptoms, particularly during adolescence and adulthood. Nevertheless, the underlying neurodevelopmental characteristics remain present and continue to influence functioning across multiple domains [1]. For a diagnosis of ASD to be established, symptoms must result in clinically significant impairment in social, occupational, educational, or other important areas of functioning. The impact of autism extends beyond the presence of diagnostic features and encompasses the ways in which these characteristics affect everyday life. Functional impairments may include challenges in academic achievement, employment, independent living, social participation, and emotional well-being. The degree of impairment varies considerably among individuals, reflecting the heterogeneous nature of the disorder. Consequently, assessment processes must consider not only symptom presentation but also the practical implications of these symptoms within real-world contexts [1].

The DSM-5-TR also specifies that ASD cannot be more appropriately explained by intellectual developmental disorder or global developmental delay. However, intellectual disability frequently co-occurs with autism, creating a complex clinical profile that requires careful diagnostic evaluation. In cases where both conditions are present, social communication deficits must exceed those expected based on the individual's overall developmental level. This distinction is essential for ensuring diagnostic accuracy and guiding intervention planning. Recognition of co-occurring conditions further contributes to a more comprehensive understanding of individual strengths, needs, and support requirements [1]. One of the notable advancements introduced by the DSM-5-TR involves the incorporation of diagnostic specifiers that allow clinicians to provide a more detailed and individualized characterization of ASD. These specifiers address factors such as symptom severity, intellectual functioning, language abilities, associated genetic or medical conditions, environmental influences, and the presence of comorbid catatonia. By incorporating these dimensions into the diagnostic process, clinicians can communicate a more nuanced description of an individual's presentation, thereby supporting personalized intervention planning and facilitating interdisciplinary collaboration. This approach reflects a broader recognition of autism as a spectrum condition characterized by substantial diversity rather than a uniform clinical entity [1]. The DSM-5-TR diagnostic framework also replaced several previously distinct diagnostic categories, including autistic disorder, Asperger disorder, and pervasive developmental disorder not otherwise specified, which were separately recognized in earlier editions of the Diagnostic and Statistical Manual of Mental Disorders. This transition toward a unified spectrum-based classification reflects accumulating evidence indicating substantial overlap among these conditions. The spectrum model acknowledges the continuous variation observed in symptom presentation and functional outcomes, promoting a more flexible and inclusive understanding of autism [1].

Contemporary perspectives on ASD increasingly recognize the broader concept of neurodiversity. According to the Centers for Disease Control and Prevention, approximately 15% to 20% of the global population demonstrates some form of neurodivergence [2]. Neurodiversity refers to the natural variation in human neurological functioning and cognitive processing, emphasizing that differences in thinking, learning, and perceiving the world are inherent aspects of human diversity rather than inherently pathological conditions [3]. Within this framework, conditions such as ASD, attention-deficit/hyperactivity disorder, and learning disabilities are viewed as naturally occurring variations in neurocognitive functioning rather than solely as deficits requiring correction. This perspective has contributed to significant shifts in public discourse, clinical practice, and advocacy efforts related to autism and other neurodevelopmental conditions [4]. The neurodiversity paradigm challenges traditional deficit-based approaches by emphasizing the strengths, capabilities, and unique perspectives of neurodivergent individuals. Historically, autistic individuals have frequently experienced stigmatization, marginalization, and negative societal perceptions. Such attitudes may contribute to barriers in education, employment, healthcare access, and social inclusion. By recognizing neurological diversity as a valuable aspect of human variation, the neurodiversity movement promotes acceptance,

inclusion, and respect for individual differences. This perspective encourages society to address environmental barriers and provide appropriate accommodations rather than focusing exclusively on attempts to normalize behavior [4]. Language preferences represent another important consideration within autism care and research. While healthcare professionals have traditionally employed person-first language, such as “individual with autism,” many autistic adults express a preference for identity-first language, including terms such as “autistic person.” These preferences reflect differing perspectives regarding the relationship between autism and personal identity. Respecting an individual's preferred terminology is considered an essential component of person-centered care and demonstrates recognition of autonomy, dignity, and lived experience [5].

The impact of ASD extends beyond affected individuals and significantly influences family systems. Parents and caregivers often encounter numerous challenges associated with supporting a child diagnosed with autism. These challenges may include social isolation, emotional stress, relationship strain, caregiving demands, and financial pressures related to healthcare, education, and specialized services. Families frequently navigate complex service systems while adapting to the unique needs of their children. Research has also demonstrated elevated recurrence rates of ASD among siblings, particularly among monozygotic twins, highlighting the substantial genetic contribution to the condition [6]. Understanding the experiences of families is therefore critical for developing comprehensive support systems that address both individual and familial needs. Despite increasing awareness and advances in diagnostic practices, significant barriers to healthcare access and service utilization persist for autistic individuals. Healthcare environments are often characterized by sensory stimuli, communication demands, and procedural uncertainties that can create substantial challenges for this population. Consequently, clinicians require structured frameworks to facilitate effective and compassionate care. One such framework is the SPACE model, which emphasizes sensory needs, predictability, acceptance, communication, and empathy as fundamental principles for supporting autistic individuals [7]. The sensory component of the SPACE framework recognizes the prevalence of sensory sensitivities among autistic individuals and the profound impact these sensitivities can have on healthcare experiences. Excessive sensory stimulation within clinical environments may result in distress, sensory overload, shutdowns, or meltdowns. The predictability component emphasizes the importance of routine, structure, and preparation, acknowledging that unexpected changes can generate significant anxiety. Acceptance focuses on reducing stigma and fostering understanding through individualized accommodations and respectful interactions. Communication highlights the need for flexible communication approaches, including augmentative and alternative communication methods when verbal communication becomes difficult during periods of stress. Finally, empathy encourages healthcare professionals to appreciate and understand the perspectives and experiences of autistic individuals, recognizing that emotional responses may differ from conventional expectations while remaining entirely valid and meaningful [7]. Collectively, these perspectives illustrate the complexity of Autism Spectrum Disorder as both a neurodevelopmental condition and a lived human experience. Contemporary approaches increasingly emphasize individualized assessment, evidence-based intervention, neurodiversity-informed practice, and person-centered care. Such approaches recognize the diverse strengths, challenges, and support needs of autistic individuals while promoting inclusion, dignity, and meaningful participation across all aspects of life.

### **Etiology**

The etiology of Autism Spectrum Disorder (ASD) remains one of the most extensively investigated areas within developmental neuroscience and psychiatric research. Contemporary evidence suggests that ASD is a multifactorial neurodevelopmental condition resulting from a complex interaction between genetic susceptibility and environmental influences. Rather than being attributable to a single cause, ASD appears to emerge from multiple converging biological pathways that influence early brain development and neural functioning. Research conducted over the past several decades has demonstrated that genetic factors play a substantial role in ASD susceptibility; however, environmental exposures during critical developmental periods may also contribute to risk. The interplay between these factors is believed to influence neurodevelopmental trajectories, resulting in the heterogeneous clinical presentations observed among individuals with ASD [8]. Current scientific understanding indicates that several biological mechanisms may contribute to the development of ASD. Although the precise pathophysiological pathways remain incompletely understood, growing evidence suggests that immune dysregulation, mitochondrial dysfunction, oxidative stress, alterations in the gut microbiome, and hormonal imbalances may influence neurodevelopmental processes associated with autism. These mechanisms are not mutually exclusive and may interact with one another to affect brain maturation, synaptic connectivity, neuronal communication, and behavioral outcomes. The complexity of these interactions reflects the multifaceted nature of ASD and highlights the challenges associated with identifying a single etiological pathway capable of explaining all cases of the disorder [8]. Genetic factors represent one of the strongest and most consistently supported contributors to ASD risk. Evidence for a substantial genetic component originates from family-based studies, twin studies, and molecular genetic investigations. Research involving monozygotic twins has demonstrated significantly higher concordance rates for ASD compared with dizygotic twins, indicating a strong hereditary influence. Similarly, increased prevalence among siblings and other family members

further supports the role of inherited genetic factors in autism susceptibility. Current estimates suggest that approximately half of ASD risk can be attributed to heritable influences, although the remaining proportion likely reflects interactions between genetic and environmental determinants [8].

The genetic architecture of ASD is highly heterogeneous and involves numerous genes rather than a single causative mutation. Advances in genomic technologies have enabled researchers to identify hundreds of genes associated with increased autism risk. These findings have transformed the understanding of ASD from a disorder linked to isolated genetic abnormalities into a condition characterized by extensive genetic complexity. Different combinations of genetic variants may contribute to autism in different individuals, thereby accounting for the remarkable diversity observed in symptom presentation, cognitive abilities, language development, adaptive functioning, and behavioral characteristics. Consequently, ASD is increasingly viewed as a spectrum of neurodevelopmental conditions that share common clinical features while arising from diverse genetic pathways [9]. One of the most notable characteristics of ASD genetics is the considerable variability in clinical outcomes among individuals carrying similar genetic alterations. People who possess comparable gene variants may exhibit markedly different developmental profiles and symptom severity. This phenomenon highlights the influence of additional genetic, epigenetic, and environmental factors that modify the expression of risk genes. The variability in phenotypic presentation suggests that genetic susceptibility alone is insufficient to determine the precise manifestation of ASD and that multiple interacting influences contribute to individual developmental trajectories [9]. Research has identified two major categories of proteins that appear particularly relevant to ASD pathogenesis. The first category includes proteins involved in synapse formation and maintenance. Synapses are specialized junctions through which neurons communicate, and disruptions in synaptic development may impair information processing, learning, memory, and social cognition. The second category comprises proteins responsible for transcriptional regulation and chromatin remodeling. These proteins influence gene expression by regulating how genetic information is accessed and utilized within cells. Abnormalities affecting these molecular processes may alter neurodevelopmental pathways during critical periods of brain maturation, thereby increasing susceptibility to ASD [9].

In addition to inherited genetic influences, several other genetic mechanisms have been implicated in autism development. *De novo* genetic variants, which arise spontaneously rather than being inherited from parents, have been identified in many individuals with ASD. Somatic mosaicism, characterized by the presence of genetically distinct cell populations within the same individual, may also contribute to autism risk. Furthermore, copy number variations (CNVs), involving deletions or duplications of segments of genetic material, represent an important source of genetic vulnerability. It is estimated that approximately 10% of ASD cases may be associated with clinically significant CNVs. Among the most extensively studied chromosomal regions linked to autism are 16p11.2 and 15q11–13. Nevertheless, these specific alterations account for only a small proportion of total ASD cases, emphasizing the considerable genetic diversity underlying the disorder [9]. Epigenetic mechanisms have emerged as another important area of investigation in ASD research. Epigenetics refers to modifications that influence gene activity without altering the underlying DNA sequence. Genes involved in epigenetic regulation may affect an individual's susceptibility to autism by modifying patterns of gene expression during critical stages of development. Several biological pathways influenced by epigenetic processes have been implicated in ASD, including those associated with synaptic transmission, immune system regulation, ion transport, and gamma-aminobutyric acid (GABA)-ergic neurotransmission. Because epigenetic processes are responsive to both genetic and environmental influences, they may represent a key mechanism through which environmental exposures contribute to autism risk [9]. Another important observation is the substantial genetic overlap between ASD and other neuropsychiatric and neurodevelopmental disorders. Studies have demonstrated shared genetic pathways among autism, schizophrenia, attention-deficit/hyperactivity disorder (ADHD), intellectual disability, and bipolar disorder. This overlap suggests that many psychiatric and developmental conditions may arise from common biological vulnerabilities that influence brain development and neural functioning. Such findings support dimensional approaches to psychopathology and highlight the interconnected nature of neurodevelopmental and psychiatric disorders [9].

The concept of genetic modifiers provides additional insight into the remarkable heterogeneity observed in ASD. Researchers have proposed that secondary genetic variants may influence the expression and severity of autism-related traits through a “second-hit” mechanism. According to this model, an individual may inherit or develop a primary susceptibility variant that increases autism risk, while additional genetic alterations subsequently modify the clinical presentation. These secondary influences may include further copy number variations, small nucleotide repeats, insertions, deletions, or other genomic changes. The interaction between primary and secondary variants may contribute to differences in symptom severity, intellectual functioning, language development, and adaptive behavior among individuals with ASD [9].

Sex differences in autism prevalence remain another important area of etiological research. Epidemiological studies consistently report higher rates of ASD diagnosis among males than females. However, the reasons underlying this disparity remain uncertain. Several hypotheses have been proposed to explain the observed sex differences. One theory suggests that females possess biological protective mechanisms that reduce vulnerability to autism despite carrying

similar genetic risk factors. Another hypothesis proposes that males are inherently more susceptible to neurodevelopmental disruptions associated with ASD. A third explanation suggests that autism may present differently in females, resulting in underrecognition or delayed diagnosis. Emerging evidence indicates that many females develop compensatory social strategies that may mask symptoms, thereby complicating identification and contributing to lower reported prevalence rates [9]. Although genetics play a central role in ASD susceptibility, environmental and perinatal influences are also believed to contribute to risk. These factors do not independently cause autism but may interact with underlying genetic predispositions to influence neurodevelopmental outcomes. Consequently, ASD is increasingly conceptualized as a condition arising from the interaction of multiple biological and environmental determinants rather than a single causative factor [8]. One of the most extensively studied environmental controversies surrounding ASD concerns childhood vaccinations. Numerous large-scale epidemiological investigations and systematic reviews have consistently demonstrated that vaccines do not cause autism. Scientific evidence has repeatedly shown that neither vaccine components such as thimerosal or mercury-containing preservatives nor the administration of multiple vaccines, including the measles-mumps-rubella (MMR) vaccine, are associated with increased ASD risk. These findings have been replicated across diverse populations and research methodologies, providing strong evidence against a causal relationship between vaccination and autism [10][11][12].

Prenatal exposure to certain infectious diseases has been identified as a potential environmental risk factor for ASD. Maternal infections during pregnancy, particularly rubella, influenza, and cytomegalovirus infections, have been associated with increased autism risk in offspring. Maternal fever during pregnancy may also contribute to altered fetal neurodevelopment. These associations support the hypothesis that maternal immune activation during critical developmental periods may influence fetal brain development through inflammatory and immunological pathways [8]. Maternal metabolic health has also emerged as an important consideration in ASD etiology. Gestational diabetes and maternal obesity have been consistently identified as risk factors for autism. These conditions may affect fetal neurodevelopment through mechanisms involving inflammation, altered glucose metabolism, hormonal dysregulation, and oxidative stress. The increasing prevalence of metabolic disorders among women of reproductive age has therefore generated considerable interest in understanding their potential contribution to ASD risk [8]. Maternal medication exposure during pregnancy represents another area of ongoing investigation. Some studies have suggested potential associations between prenatal exposure to selective serotonin reuptake inhibitors (SSRIs) and other antidepressants and an increased likelihood of ASD. However, these associations often diminish substantially when researchers control for underlying maternal psychiatric conditions. Consequently, it remains difficult to determine whether observed risks are attributable to medication exposure itself or to the psychiatric disorders for which these medications are prescribed [8]. Research has also explored the potential influence of maternal antibiotic use during pregnancy on ASD development. Some findings suggest that prenatal antibiotic exposure may alter maternal and fetal microbiota, potentially affecting neurodevelopmental processes. Although evidence remains inconclusive, this area continues to receive attention because of growing recognition of the relationship between the gut microbiome and brain development [8].

Environmental toxicants have been widely investigated as possible contributors to ASD susceptibility. Prenatal exposure to phthalates, air pollutants, pesticides, and heavy metals such as lead and mercury has been associated with increased autism risk in some studies. These environmental agents may affect fetal development through mechanisms involving oxidative stress, endocrine disruption, immune activation, and neurotoxicity. Nevertheless, the magnitude of risk associated with individual exposures remains variable, and further research is required to clarify these relationships [8]. Additional factors that may influence ASD risk include advanced maternal age, advanced paternal age, the use of assisted reproductive technologies, maternal nutritional status during pregnancy, and prenatal exposure to medications such as valproic acid and other antiepileptic agents. These associations further illustrate the multifactorial nature of autism etiology and emphasize that ASD arises from the interaction of numerous biological and environmental influences operating across developmental stages [13]. Overall, contemporary evidence supports a multidimensional model of ASD etiology in which genetic susceptibility serves as the primary foundation upon which environmental, epigenetic, immunological, metabolic, and developmental influences act. The substantial heterogeneity observed in autism reflects the complexity of these interactions and underscores the need for continued research aimed at elucidating the biological mechanisms that contribute to ASD development. A deeper understanding of these etiological pathways may ultimately facilitate earlier identification, more precise risk assessment, and the development of targeted prevention and intervention strategies for individuals with ASD and their families.

### **Epidemiology**

The epidemiology of Autism Spectrum Disorder (ASD) has undergone substantial transformation over the past several decades, reflecting not only genuine changes in prevalence but also significant shifts in diagnostic conceptualization, classification systems, public awareness, and case identification practices. Consequently, interpreting epidemiological trends in autism requires careful consideration of the historical evolution of diagnostic criteria and the methodological differences that influence prevalence estimates across time and geographic regions. The changing definition of autism

has played a central role in shaping contemporary understanding of the disorder's prevalence and distribution within populations. The concept of autism has evolved considerably since its initial introduction in psychiatric literature. The term "autism" was first coined by Eugen Bleuler in 1911 to describe profound social withdrawal and self-focused behavior observed among individuals with schizophrenia. At that time, autism was not considered a distinct developmental disorder but rather a symptom associated with psychotic conditions. Several decades later, Leo Kanner significantly advanced the understanding of autism by describing a group of children who exhibited marked difficulties in language development, social interaction, and behavioral flexibility. Kanner subsequently emphasized extreme social isolation and a strong preference for sameness as defining characteristics of the condition, thereby establishing the foundation for modern conceptualizations of autism [14][15]. Further contributions to the field emerged through the work of Hans Asperger, who described children demonstrating social communication difficulties despite possessing average or above-average intellectual and language abilities. Asperger's observations eventually influenced the development of the diagnostic category known as Asperger syndrome. Over time, autism-related conditions were incorporated into broader diagnostic frameworks, culminating in the publication of the Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition (DSM-IV) in 1994. This classification system grouped autistic disorder, Asperger syndrome, Rett syndrome, childhood disintegrative disorder, and pervasive developmental disorder not otherwise specified under the umbrella category of pervasive developmental disorders [16]. Subsequent historical analyses, however, have revealed troubling aspects of Asperger's involvement with Nazi-era child euthanasia programs, leading to significant criticism and reassessment of his legacy within the field of child psychiatry [17].

Advances in clinical research and diagnostic understanding eventually prompted substantial revisions to autism classification systems. Concerns regarding the validity and diagnostic utility of certain pervasive developmental disorder subtypes, particularly childhood disintegrative disorder, together with recognition of Rett syndrome as a distinct genetic condition, contributed to a major reorganization of diagnostic criteria. In 2013, the DSM-5 replaced the category of pervasive developmental disorders with the unified diagnosis of Autism Spectrum Disorder. This revision eliminated previous subcategories and introduced a single diagnostic framework based on two core domains: persistent deficits in social communication and social interaction, and restricted, repetitive patterns of behavior, interests, or activities [18]. The adoption of a spectrum-based model acknowledged the considerable heterogeneity of autism and provided a more comprehensive approach to diagnosis. The evolving diagnostic framework has had profound implications for epidemiological research. Differences in prevalence estimates across studies are often influenced by variations in diagnostic criteria, assessment methods, case-finding procedures, healthcare infrastructure, and cultural attitudes toward developmental disorders. Consequently, comparisons across countries and historical periods must be interpreted cautiously. The prevalence of ASD is not determined solely by biological factors but is also shaped by the capacity of healthcare systems to identify affected individuals, the availability of diagnostic services, public awareness of autism, and the inclusiveness of surveillance methods [19]. Historical prevalence studies illustrate the dramatic changes that have occurred in autism identification over time. One of the earliest epidemiological investigations was conducted in England and published in 1967. This study examined children aged eight to ten years and reported a prevalence estimate of approximately 4.5 cases per 10,000 children for what was then described as "autistic conditions of early childhood" [20]. Such estimates were substantially lower than contemporary figures, reflecting narrower diagnostic definitions, limited awareness among clinicians and educators, and restricted access to specialized assessment services. Many individuals who would meet current diagnostic criteria for ASD were likely unrecognized or classified under alternative developmental or psychiatric diagnoses.

Recent epidemiological evidence demonstrates a substantial increase in reported autism prevalence worldwide. A comprehensive global meta-analysis revealed significant regional variation in ASD prevalence, with pooled estimates of approximately 1.01% in North America, 0.73% in Europe, and 0.41% in Asia [21]. The highest reported prevalence rates were observed in the United States, where approximately 1.12% of the population was estimated to have ASD, followed by Sweden at 0.90% and Denmark at 0.73%. In contrast, lower prevalence estimates were reported in Taiwan at 0.11%, France at 0.32%, and China at 0.42% [21]. These differences likely reflect a combination of methodological factors, cultural influences, healthcare accessibility, and variations in diagnostic practices rather than true biological differences in autism occurrence across populations. Global prevalence trends indicate a marked increase in ASD identification over recent decades. Data from international studies demonstrate that the estimated point prevalence of ASD rose from approximately 0.25% during the period between 1994 and 1999 to nearly 0.99% between 2015 and 2019 [21]. Several explanations have been proposed for this increase. Expanded diagnostic criteria, improved awareness among healthcare professionals and the public, enhanced screening initiatives, earlier identification practices, and greater access to specialized services have all contributed to rising prevalence estimates. Although some researchers continue to investigate whether environmental and societal factors may also play a role, much of the observed increase is generally attributed to changes in recognition and diagnosis rather than a true epidemic increase in disease occurrence. In the United States, surveillance data from the Centers for Disease Control and Prevention (CDC) have provided some of the most comprehensive epidemiological information regarding ASD. According to CDC estimates, approximately one in every thirty-six children aged eight years met diagnostic criteria for ASD in

2020 [2]. These findings highlight autism as one of the most common neurodevelopmental conditions affecting children. The increasing prevalence identified through national surveillance programs underscores the growing need for healthcare, educational, and social support services designed to address the diverse needs of autistic individuals and their families.

Sex differences remain one of the most consistently observed epidemiological characteristics of ASD. Current data indicate that autism is diagnosed considerably more frequently in males than in females. According to CDC estimates, approximately 4% of boys and 1% of girls in the United States are diagnosed with ASD [2]. However, growing evidence suggests that these figures may not fully reflect the true prevalence of autism among females. Researchers increasingly recognize that girls and women may present with different symptom profiles, exhibit stronger compensatory social behaviors, and develop masking strategies that obscure autistic characteristics during clinical evaluations. As a result, many females may remain undiagnosed, receive alternative psychiatric diagnoses, or obtain an autism diagnosis later in life. The recognition of sex-related differences in autism presentation has contributed to changing diagnostic practices and increased awareness among clinicians. Epidemiological data indicate that the proportion of girls diagnosed with ASD has steadily increased between 2000 and 2021 [22]. This trend likely reflects improvements in understanding how autism manifests in females rather than a true increase in incidence among girls. Enhanced awareness of female-specific presentations has encouraged more comprehensive assessment approaches and reduced the likelihood that autistic females will be overlooked during diagnostic evaluations. Racial and ethnic disparities in ASD identification have also been an important focus of epidemiological research. Historically, autism prevalence appeared substantially higher among White children than among Black or Hispanic children. Earlier estimates suggested that ASD prevalence among White children exceeded that of Black and Hispanic children by approximately 50% [2]. However, growing evidence indicates that these disparities were largely influenced by inequities in healthcare access, diagnostic opportunities, socioeconomic factors, and differences in service utilization. As autism awareness has expanded and diagnostic services have become more accessible across diverse populations, the gap in prevalence estimates among racial and ethnic groups has narrowed considerably [2].

The reduction in racial and ethnic disparities represents an important achievement in public health and developmental healthcare. More equitable identification practices ensure that children from diverse backgrounds have improved access to diagnostic assessments, early intervention services, educational accommodations, and family support programs. Nevertheless, ongoing efforts remain necessary to address persistent barriers to diagnosis and treatment, particularly among underserved and marginalized communities. Overall, the epidemiology of Autism Spectrum Disorder reflects a dynamic interaction between evolving diagnostic frameworks, increased public and professional awareness, improved surveillance systems, and broader access to healthcare services. The substantial rise in prevalence observed over recent decades is best understood within the context of these developments rather than as evidence of a singular causal phenomenon. Contemporary epidemiological research continues to enhance understanding of the global distribution of ASD while highlighting the importance of equitable identification, culturally sensitive assessment practices, and comprehensive support systems for autistic individuals across the lifespan [2][21].

### **Pathophysiology**

Despite decades of extensive scientific investigation, the pathophysiology of Autism Spectrum Disorder (ASD) remains incompletely understood. Current evidence suggests that ASD arises from complex interactions among genetic, neurobiological, and environmental factors that influence brain development during critical prenatal and early postnatal periods. Although no single biological mechanism has been identified as responsible for all cases of autism, numerous studies support the view that abnormalities in neural connectivity and synaptic development play central roles in the disorder's underlying pathogenesis. ASD is therefore increasingly conceptualized as a disorder of atypical brain development characterized by alterations in neuronal organization, communication, and functional integration across multiple brain regions [23]. Among the most widely supported theories is the hypothesis of impaired neural connectivity and disrupted synaptogenesis. Synaptogenesis refers to the formation and refinement of synaptic connections between neurons, a process that is essential for normal cognitive, social, and behavioral functioning. Research suggests that individuals with ASD may exhibit an increased number of neurons and atypical synaptic connections during early brain development. These abnormalities may interfere with the normal pruning and fine-tuning of neural circuits, leading to inefficient communication between different brain regions. Such disruptions may contribute to the social communication difficulties, repetitive behaviors, and sensory processing differences commonly observed in ASD [23]. Several additional neurodevelopmental theories have been proposed to explain the biological basis of autism. These include abnormalities in neuronal migration, altered dendritic growth and morphogenesis, and imbalances between excitatory and inhibitory neurotransmission within neural networks. The excitation-inhibition imbalance hypothesis proposes that excessive excitatory signaling or insufficient inhibitory control may disrupt information processing and neural synchronization, thereby contributing to autistic behaviors. Other theories focus on dysfunction within the mirror neuron system, which is believed to support social cognition,

imitation, and understanding the intentions and emotions of others. Furthermore, increasing evidence suggests that immune dysregulation, chronic neuroinflammation, and epigenetic modifications may influence brain development and contribute to ASD susceptibility and symptom expression [23].

Neuroimaging research has provided valuable insights into the neurobiological characteristics of ASD. Studies using structural and functional brain imaging techniques have identified differences in brain volume, gray matter distribution, white matter integrity, and neural connectivity patterns among autistic individuals. However, no single neuroanatomical signature has been consistently identified across all individuals with ASD. This variability reflects the heterogeneous nature of the disorder and suggests that multiple neurobiological pathways may lead to similar clinical manifestations. One of the most consistently observed findings is an atypical developmental trajectory characterized by accelerated brain growth during early childhood followed by a period of slowed growth or relative regression later in development [24]. Environmental influences are also believed to contribute substantially to ASD risk and may account for a significant proportion of variability in autism susceptibility. Current estimates suggest that environmental factors may explain up to 50% of the risk variance associated with ASD [25]. Factors that have been associated with increased risk include advanced maternal or paternal age, prematurity, birth complications, and prenatal exposure to environmental toxicants. Exposure to heavy metals, certain medications, air pollutants, and endocrine-disrupting chemicals during critical developmental periods may interact with genetic vulnerabilities and alter neurodevelopmental processes. These findings support the contemporary understanding of ASD as a multifactorial condition resulting from complex interactions between biological predisposition and environmental influences rather than a single causative mechanism [25].

### **History and Physical**

The American Academy of Pediatrics (AAP) revised its clinical guidance in 2020 in the report titled “The Identification, Evaluation, and Management of Children with Autism Spectrum Disorder,” aligning recommendations with contemporary evidence-based practice. The updated guidance emphasizes early identification and systematic screening of all children for Autism Spectrum Disorder (ASD), recognizing that early detection and intervention significantly influence developmental trajectories and long-term functional outcomes. Screening programs are primarily designed to detect early indicators of developmental delay, particularly in language acquisition, cognitive functioning, and motor development, which may signal the need for further diagnostic evaluation [6][26]. The diagnostic process for ASD is fundamentally clinical and is based on the identification of characteristic behavioral features as defined in the DSM-5-TR criteria. A definitive diagnosis requires a trained clinician who is capable of integrating detailed developmental history with direct behavioral observation. This combined approach enables a comprehensive understanding of the individual’s developmental profile, symptom presentation, and functional impairments. Clinical assessment is not based on a single test but rather on a synthesis of historical, observational, and standardized diagnostic information [6].

A thorough evaluation for suspected ASD includes multiple components. A comprehensive developmental, medical, social, and family history is essential, with particular attention to prenatal, perinatal, and postnatal risk factors that may influence neurodevelopment. These include prematurity, exposure to environmental toxins or teratogenic substances, traumatic experiences, and a family history of developmental or psychiatric disorders. A complete physical examination is also required, including measurement of growth parameters, neurological assessment, and inspection for dysmorphic features or cutaneous signs that may indicate underlying genetic or neurocutaneous conditions. Sensory evaluation, including formal hearing and vision testing, is necessary to exclude sensory impairments that may mimic or contribute to developmental delays [6]. Structured behavioral observation is a critical component of the diagnostic process and requires clinical expertise in developmental assessment. Standardized tools and validated questionnaires are frequently used to support clinical evaluation and improve diagnostic accuracy. Instruments such as the Social Communication Questionnaire and the Social Responsiveness Scale assist in systematically documenting symptoms related to social communication deficits and restricted or repetitive behaviors. In research and specialized clinical contexts, the Autism Diagnostic Interview-Revised is used as a comprehensive caregiver interview to obtain detailed developmental history and behavioral information. Additionally, standardized observational measures such as the Autism Diagnostic Observation Schedule, Second Edition (ADOS-2), and the Childhood Autism Rating Scale, Second Edition, provide structured frameworks for assessing autism-related behaviors. The ADOS-2, in particular, is widely regarded as a gold-standard tool but requires specialized training for accurate administration and interpretation [6].

Early identification of ASD is often guided by the presence of developmental “red flags” that emerge during infancy and early childhood. These include failure to respond to one’s name by 12 months of age, lack of pointing or gesturing to express interest by 14 months, and absence of pretend play behaviors by 18 months. Additional early warning signs include reduced eye contact, preference for solitary activities, difficulties in understanding or expressing emotions, delayed speech and language development, echolalia or repetitive use of language, irrelevant responses to questions, and distress in response to minor changes in routine. Other characteristic features may include restricted or highly

focused interests, repetitive motor behaviors, and atypical responses to sensory stimuli, such as heightened sensitivity or reduced reactivity to environmental input. Recognition of these early indicators is essential for prompt referral, comprehensive assessment, and initiation of early intervention services [6].

### **Evaluation**

The evaluation of Autism Spectrum Disorder (ASD) is primarily clinical, as there are no specific laboratory investigations or neuroimaging modalities that can independently confirm the diagnosis. Diagnosis is established through comprehensive clinical assessment based on behavioral criteria outlined in the DSM-5-TR, supported by developmental history, caregiver reports, and structured observation. Laboratory and imaging studies are not used as diagnostic tools for ASD itself but may be indicated to identify underlying or associated medical conditions that can present with similar features or contribute to neurodevelopmental abnormalities [6]. In selected cases, referral for pediatric genetic evaluation is recommended to investigate potential syndromic or hereditary conditions associated with autistic features. Genetic assessment may provide insight into underlying chromosomal or single-gene disorders, particularly in individuals with dysmorphic features, intellectual disability, or a strong family history of neurodevelopmental disorders. Identifying a genetic etiology can also assist in prognosis, counseling, and long-term management planning [6]. When clinically indicated, additional metabolic and biochemical investigations may be considered to rule out inborn errors of metabolism or other systemic conditions that can affect neurological development. These may include plasma amino acid levels, urine organic acid analysis, and acylcarnitine profiling. Such investigations help detect metabolic disorders that may present with developmental delay or regression. Liver and thyroid function tests may also be performed to exclude systemic endocrine or hepatic dysfunction that could contribute to neurodevelopmental impairment. Measurement of blood lead levels is important in cases where environmental exposure to neurotoxic substances is suspected, given the established impact of lead toxicity on cognitive and behavioral development [6].

Neuroimaging studies are not routinely required in the evaluation of ASD but may be indicated in specific clinical scenarios. Brain imaging, such as magnetic resonance imaging (MRI), should be considered in individuals presenting with atypical developmental regression, microcephaly or macrocephaly, seizures, abnormal neurological examination findings, or cranial structural abnormalities. These findings may suggest underlying neurological pathology that warrants further investigation beyond the diagnosis of ASD [6]. Electroencephalography (EEG) may also be utilized in cases where seizure activity is suspected or when there is unexplained developmental regression, particularly loss of previously acquired language skills. Additional neurological symptoms, such as episodic behavioral changes or altered consciousness, may also justify EEG assessment. Although EEG abnormalities are not specific to ASD, they may indicate coexisting epileptic disorders or other neurological conditions that require targeted management [6]. Overall, the evaluation of ASD involves a judicious and individualized approach, focusing on clinical diagnosis while selectively employing laboratory, genetic, and neuroimaging studies to exclude alternative or comorbid conditions. This comprehensive strategy ensures accurate diagnosis, appropriate identification of underlying etiologies, and optimized patient management [6].

### **Treatment / Management**

Autism Spectrum Disorder (ASD) is a lifelong neurodevelopmental condition that requires an individualized, multidisciplinary, and developmentally informed approach to management. The heterogeneity of ASD necessitates tailoring interventions according to the individual's age, cognitive abilities, language level, adaptive functioning, and behavioral profile. Management strategies must also consider personal strengths and vulnerabilities, recognizing that autistic individuals may present with both significant challenges and unique abilities. Contemporary perspectives increasingly emphasize a neurodiversity-informed approach, in which autism is not only viewed as a disability requiring support but also as a variation in human neurodevelopment that may include distinct cognitive strengths and abilities. This dual perspective supports interventions that enhance quality of life while respecting individual identity and autonomy [4]. In many countries, particularly the United States, legal frameworks have been established to ensure access to appropriate educational and therapeutic services for individuals with developmental disabilities, including ASD. Policies such as the No Child Left Behind Act of 2001, the Individuals with Disabilities Education Improvement Act of 2004, and the Every Student Succeeds Act of 2015 provide a structured legal foundation for delivering free and evidence-based educational interventions. These laws ensure that children with ASD receive individualized educational planning, early intervention services, and accommodations that support academic participation and developmental progress within inclusive learning environments. Evidence-based management of ASD is broadly categorized into focused intervention practices and comprehensive treatment models. Focused intervention practices target specific skills or behavioral outcomes, while comprehensive programs provide structured, long-term intervention frameworks that address multiple developmental domains simultaneously. According to evidence-based reviews, these interventions are delivered across a range of settings including early intervention programs, schools, community organizations, and healthcare systems. Effective intervention programs are characterized by individualized

assessment, goal setting, interdisciplinary collaboration, trained professionals, structured learning environments, behavioral support strategies, and active family involvement. Continuous monitoring of progress and transition planning are also essential components of high-quality care [6][27].

A wide range of evidence-based focused intervention practices has been identified for individuals with ASD, each targeting specific developmental and behavioral outcomes. Behavioral interventions such as antecedent-based strategies, reinforcement procedures, and extinction techniques are commonly used to reduce challenging behaviors and promote adaptive functioning. Communication-focused interventions, including augmentative and alternative communication systems and functional communication training, are essential for individuals with limited verbal abilities. Instructional approaches such as discrete trial training and direct instruction are widely applied to develop academic, cognitive, and language skills in structured learning environments. Naturalistic developmental behavioral interventions emphasize learning within everyday contexts and social interactions, promoting generalization of skills across environments. Parent-implemented interventions play a critical role in extending therapeutic strategies into the home environment, enhancing consistency and developmental progress. Peer-mediated approaches facilitate social interaction and communication skills through structured engagement with typically developing peers. Additional interventions such as modeling, video modeling, and social narratives support learning through visual and observational methods, which are often particularly effective for autistic learners. Other important intervention strategies include social skills training, sensory integration approaches, self-management techniques, and task analysis for breaking complex skills into manageable steps. Technology-aided interventions, including digital learning tools and assistive communication devices, are increasingly utilized to support communication, learning, and independence. Time delay procedures, prompting strategies, and reinforcement systems are also widely applied to facilitate skill acquisition and behavioral regulation. Collectively, these interventions address a broad range of developmental domains including communication, social interaction, academic skills, adaptive functioning, and vocational preparation [27].

The effectiveness of ASD interventions depends heavily on implementation in real-world settings by trained professionals such as teachers, speech-language pathologists, psychologists, and behavioral therapists. Ongoing evaluation of intervention outcomes in naturalistic environments is essential for determining effectiveness and ensuring that evidence-based practices are appropriately translated into routine clinical and educational services. Future research continues to emphasize the importance of ecological validity in intervention studies to ensure that outcomes are meaningful and applicable to everyday functioning [27]. Pharmacological treatment does not target the core symptoms of ASD, such as social communication deficits or restricted and repetitive behaviors. Instead, medication is primarily used to manage associated behavioral, emotional, and psychiatric comorbidities. Clinical assessment must first exclude physical sources of distress, such as pain, gastrointestinal disorders, infections, or sensory discomfort, which may contribute to behavioral changes. In addition, psychiatric comorbidities are highly prevalent among individuals with ASD, with conditions such as attention-deficit/hyperactivity disorder, anxiety disorders, obsessive-compulsive disorder, mood disorders, and conduct-related disorders occurring in a substantial proportion of children and adolescents. Estimates suggest that between 70% and 90% of individuals with ASD experience at least one co-occurring psychiatric condition, highlighting the importance of comprehensive mental health assessment [6]. When behavioral and environmental interventions are insufficient to manage clinically significant symptoms such as severe aggression, self-injurious behavior, sleep disturbances, or severe emotional dysregulation, pharmacological interventions may be considered. Medication selection is guided by symptom profile rather than ASD itself. For symptoms of hyperactivity, impulsivity, and inattention, treatment options may include psychostimulants, atomoxetine, clonidine, guanfacine, and atypical antipsychotic medications. In cases of severe irritability, aggression, or self-injurious behavior, atypical antipsychotics are commonly used, alongside agents such as clonidine, guanfacine, selective serotonin reuptake inhibitors, valproic acid, and venlafaxine.

For repetitive behaviors, atypical antipsychotics, valproic acid, and serotonin reuptake inhibitors may be considered, depending on symptom severity and functional impact. Anxiety and depressive symptoms may be managed using serotonin reuptake inhibitors, clonidine, guanfacine, and atypical antipsychotic medications when clinically indicated. Medication management must always be individualized, carefully monitored, and integrated within a broader therapeutic plan that includes behavioral, educational, and psychosocial interventions. The use of psychotropic medications in ASD requires a careful balance between potential benefits and risks. Informed consent from caregivers and, when appropriate, the patient is essential prior to initiating treatment. Continuous monitoring for efficacy, side effects, and functional outcomes is necessary to ensure safe and appropriate use. Pharmacological therapy should never be used in isolation but should form part of a comprehensive, multidisciplinary approach aimed at improving quality of life, reducing symptom burden, and supporting functional independence [6].

### **Differential Diagnosis**

Autism Spectrum Disorder (ASD) requires careful diagnostic evaluation because its core features, particularly social communication difficulties and repetitive behaviors, overlap with a wide range of genetic, neurological, metabolic,

and psychiatric conditions. Accurate differentiation is essential to avoid misdiagnosis and to ensure appropriate management. A major group of differential diagnoses includes genetic syndromes associated with developmental delay and behavioral abnormalities. These include tuberous sclerosis complex, which presents with multisystem tumors, seizures, and cognitive impairment, and Fragile X syndrome, commonly linked to intellectual disability and characteristic physical features. Other chromosomal and genetic disorders such as 15q11–q13 duplication syndrome, Angelman syndrome, Rett syndrome, and Down syndrome may present with language impairment, motor abnormalities, or severe developmental delay resembling ASD. Additional syndromes such as Neurofibromatosis type 1, Noonan syndrome, Williams-Beuren syndrome, DiGeorge syndrome, Smith-Lemli-Opitz syndrome, Timothy syndrome, and Joubert syndrome may also mimic autistic features through a combination of neurological, cardiac, or developmental abnormalities [6][28]. Environmental and metabolic conditions must also be excluded during assessment. These include heavy metal toxicity such as lead or mercury exposure, aminoacidopathies, hypothyroidism, organophosphate poisoning, and seizure disorders, particularly those presenting with atypical or subtle seizure activity [6]. Several psychiatric disorders can closely resemble or coexist with ASD. According to DSM-5-TR, these include attention-deficit/hyperactivity disorder, intellectual developmental disorder, language disorders, social (pragmatic) communication disorder, selective mutism, stereotypic movement disorder, anxiety disorders, obsessive-compulsive disorder, schizophrenia, mood disorders, and personality disorders. These conditions may share behavioral or communicative impairments, making differential diagnosis complex and clinically significant. Rare conditions such as glycogen storage diseases and autoimmune encephalitis may also present with developmental regression or behavioral changes that resemble ASD [29][30]. Overall, distinguishing ASD from these conditions requires comprehensive clinical history, developmental assessment, neurological examination, and targeted investigations. A holistic evaluation ensures diagnostic accuracy and appropriate treatment planning.

### **Prognosis**

The prognosis of Autism Spectrum Disorder (ASD) is highly variable and cannot be accurately predicted at the time of diagnosis. Clinical outcomes differ widely depending on individual developmental profiles, cognitive functioning, and access to intervention services. Although some children demonstrate meaningful improvement in social communication and adaptive functioning over time, longitudinal observations indicate that the majority of young children diagnosed with ASD continue to meet diagnostic criteria beyond early childhood, particularly when symptoms are moderate to severe [31]. Prognostic outcomes are more favorable in individuals who demonstrate higher cognitive abilities at an early age, stronger verbal intelligence, reduced repetitive behaviors, and earlier engagement with structured intervention programs. The availability of supportive family environments and community-based resources also contributes positively to developmental progress [6]. In contrast, deficits in executive functioning are associated with poorer long-term outcomes, particularly in adaptive behavior and independence. ASD is also associated with increased risk of premature mortality, including elevated risk of suicide, emphasizing the need for long-term monitoring and comprehensive psychosocial support [32][33].

### **Complications**

Individuals with Autism Spectrum Disorder (ASD) experience a high rate of medical and psychiatric comorbidities, which significantly contribute to disease burden and functional impairment. Clinical studies indicate that approximately 74% of individuals with ASD present with at least one co-occurring condition [34]. These comorbidities often complicate diagnosis and management because individuals with ASD may present with atypical or non-specific symptom patterns, making recognition of additional disorders more challenging. Common associated conditions include sleep disturbances, epilepsy, macrocephaly, hydrocephalus, cerebral palsy, migraines, and other neurological abnormalities. Developmental and systemic issues such as obesity, gastrointestinal disturbances, enuresis, and encopresis are also frequently reported. Psychiatric comorbidities including attention-deficit/hyperactivity disorder, anxiety disorders, mood disorders, and behavioral disturbances are highly prevalent. A significant proportion of individuals with ASD also experience intellectual disability and communication limitations, with approximately 30% being minimally verbal [6][34][35]. Behavioral complications such as aggression, self-injury, wandering, and severe tantrums further increase caregiver burden and negatively affect long-term functional outcomes.

### **Consultations**

Management of Autism Spectrum Disorder (ASD) often requires multidisciplinary collaboration due to its complex clinical presentation and frequent comorbidities. Referral for pediatric genetic evaluation is recommended to identify potential underlying syndromic or hereditary conditions [6]. ASD commonly coexists with multiple medical and neurological issues, including motor abnormalities, gastrointestinal disorders, epilepsy, intellectual disability, and sleep disturbances, necessitating involvement of various specialties. Motor abnormalities are observed in approximately 79% of individuals, gastrointestinal symptoms up to 70%, epilepsy up to 30%, intellectual disability in about 45%, and sleep disorders in 50% to 80% of cases [36]. Depending on clinical presentation, referrals may include

neurology for seizure management, gastroenterology for digestive concerns, and sleep medicine or pulmonology for sleep-related disorders. Ongoing follow-up with developmental pediatricians, child neurologists, psychologists, and psychiatrists is essential. These specialists provide continuous monitoring, behavioral management strategies, and targeted interventions to address evolving medical, developmental, and psychological needs across the lifespan.

### Patient Education

Effective care for individuals with Autism Spectrum Disorder (ASD) depends strongly on coordinated healthcare delivery and comprehensive patient and family education. Many individuals with ASD face barriers in accessing integrated services, which can negatively impact long-term outcomes [6]. Families often experience significant psychological, social, and financial stress, including social isolation, emotional strain, and difficulties navigating healthcare and educational systems. A structured, family-centered approach such as the medical home model is recommended, where primary care providers coordinate care and collaborate closely with families in decision-making and long-term planning. This model strengthens communication between clinicians and families and supports individualized care planning [6]. Shared decision-making is essential, particularly in situations where evidence-based consensus is limited, ensuring that caregivers and individuals with ASD are actively involved in care choices. Education should also focus on enhancing parental coping strategies, communication skills, and behavioral management techniques, while encouraging peer support, self-care, and advocacy. Clinicians play a key role in building trust and strengthening family resilience [37][38].

### Conclusion

Autism Spectrum Disorder represents a complex neurodevelopmental condition characterized by persistent deficits in social communication and restricted, repetitive behaviors. The disorder demonstrates marked heterogeneity in clinical presentation, etiology, and outcomes, reflecting interactions between genetic, environmental, neurobiological, and epigenetic factors. Advances in diagnostic frameworks, particularly DSM-5-TR, have improved case identification through standardized criteria and specifiers that capture severity and functional impairment. Early detection and intervention remain critical determinants of prognosis, with evidence supporting improved developmental trajectories when support is initiated in early childhood. Management requires a multidisciplinary, individualized approach that integrates behavioral, educational, psychological, and pharmacological strategies tailored to specific needs. Despite progress in understanding ASD, challenges persist in differential diagnosis, access to services, and long-term care coordination. Increasing recognition of neurodiversity has also shifted perspectives toward inclusion, acceptance, and strength-based models of care. Continued research is essential to clarify underlying mechanisms and enhance targeted interventions that improve quality of life for individuals with ASD and their families.

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