Comprehensive Review of Autism Spectrum Disorder: Etiology, Early Signs, and Diagnostic Assessment

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Abstract: Autism Spectrum Disorder (ASD) is a complex neurodevelopmental disorder characterized by challenges in social communication and interaction, restricted interests, and repetitive behaviours. This review article provides an extensive examination of ASD, covering its etiology, early signs and symptoms, and diagnostic assessment. By synthesizing current research findings, this review aims to enhance understanding of ASD and facilitate early detection, accurate diagnosis, and effective interventions.

Keywords: Autism Spectrum Disorder, etiology, early signs, symptoms, diagnostic assessment, neurodevelopmental disorder, social communication, repetitive behaviours.

1. Introduction

Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder characterized by difficulties in social communication and interaction, as well as restricted interests and repetitive behaviours (1). It affects individuals across various age groups, impacting their daily functioning and overall quality of life. Early recognition and accurate diagnosis of ASD are crucial for initiating appropriate interventions and support. Paediatricians play a vital role in the early identification of ASD, as they are often the first point of contact for parents. In recent years, increased awareness and understanding of ASD, along with advancements in diagnostic tools and assessment approaches, have improved the early detection and diagnosis of the disorder. This comprehensive review article aims to provide an in-depth exploration of the etiology of ASD, early signs and symptoms, and diagnostic assessment approaches. By synthesizing the available data and research findings, this review aims to enhance our understanding of ASD and contribute to early detection, accurate diagnosis, and effective interventions for individuals with ASD.

Definition and Diagnostic Criteria for Autism Spectrum Disorder

Autism Spectrum Disorder (ASD) is diagnosed based on a set of criteria outlined in the Diagnostic and Statistical Manual of Mental Disorders (DSM-5) (2)(3)(4)(5)(6)(7). To receive an ASD diagnosis, individuals must meet specific criteria related to social communication and interaction, as well as restricted and repetitive behaviors. The diagnostic criteria include:

1) Difficulties in Social Emotional Reciprocity

Individuals with ASD exhibit challenges in social interaction, such as trouble with social approach, engaging in back - and - forth conversations, sharing interests with others, and expressing or understanding emotions. They may have difficulty initiating or sustaining social interactions and struggle with reciprocal communication.

2) Difficulties in Nonverbal Communication

Individuals with ASD may demonstrate difficulties in nonverbal communication used for social interaction. This includes abnormal eye contact, atypical body language, and difficulty understanding and using nonverbal cues like facial expressions or gestures for communication.

3) Deficits in Developing and Maintaining Relationships

ASD is characterized by deficits in developing and maintaining relationships with others, beyond those with caregivers. Individuals with ASD may lack interest in forming social connections, have difficulties responding appropriately in different social contexts, and struggle with sharing imaginative play with peers.

In addition to the above criteria, the DSM-5 also specifies that individuals must exhibit at least two of the following four types of restricted and repetitive behaviors, interests, or activities:

• Stereotyped Speech and Repetitive Movements: This includes repeating words or phrases, engaging in repetitive motor movements, using objects or phrases in a repetitive manner, or displaying echolalia.

• Rigidity and Resistance to Change: Individuals with ASD often demonstrate a rigid adherence to routines, rituals, or patterns of behavior. They may have an extreme resistance to change and become significantly distressed when faced with even minor disruptions to their established routines.

• Highly Restricted Interests: People with ASD may exhibit a strong attachment to unusual objects or develop intense, focused interests in specific topics or activities. These interests often surpass what is considered typical for age-appropriate.

• Sensory Reactivity: Individuals with ASD may display increased or decreased reactivity to sensory input. This can manifest as a lack of response to pain, a strong dislike or sensitivity to certain sounds, excessive touching or smelling of objects, or a fascination with spinning objects.

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By using the DSM - 5 criteria (8), clinicians can maximize diagnostic sensitivity and specificity, especially in preschool children who may present with early signs of ASD. Globally, the prevalence of autism varies, with studies reporting a range of estimates (9). However, the median prevalence is approximately 100 cases per 10,000 individuals, with a male - to - female ratio of around 4.2. (10) (11) (12), (13), (14). Many individuals with ASD face challenges in communication, social interaction, and functioning effectively in various areas of life. This places significant burdens on parents and caregivers, who often experience decreased parenting efficacy, increased stress, and higher rates of mental and physical health problems compared to parents of typically developing children or children with other developmental disorders (15).

Early Signs and Symptoms: Indicators of Abnormal Brain Development

Autism Spectrum Disorder (ASD) encompasses a range of related disorders characterized by impairments in reciprocal social interaction, communication, and the presence of repetitive and inflexible behavior (16). The early symptoms of ASD can manifest in the first few months of life, providing valuable clues about atypical developmental trajectories and potential challenges ahead. (17)(18). These early signs and symptoms include delayed speech and language development or a lack of responsiveness when the Child’s name is called. Other notable differences may involve social orienting, joint attention(19) (the ability to share attention with others), imitation, affect regulation, increased negative affect, ambiguous affective expressions, and reduced use of gestures(20)(21).

Various domains of development can exhibit atypicalities in individuals with ASD. In the visual domain, there may be abnormalities in visual tracking, prolonged fixation on objects (22), and intense visual inspection(23). Motor skills may be delayed, with decreased activity levels, delayed fine and gross motor skills, and the presence of atypical motor mannerisms (24) Play behaviors may also demonstrate delays, such as limited toy play, delays in motor imitation, and repetitive actions with toys. Social - communication abilities may be affected, presenting as atypical eye gaze, difficulties in orienting to one’s name, impaired imitation, reduced social smiling, diminished social interest and affect, and a reduced expression of positive emotions (25). Language development may be delayed, including delays in babbling, particularly in back - and - forth social babbling, as well as delays in verbal comprehension and expression, and gesturing, as assessed through standardized measures (26). General cognitive development may also show signs of slower acquisition of new skills, as indicated by declining standard scores in certain domains (27), in a subset of toddlers subsequently diagnosed with ASD. [(23), (25), (27), (28), (29), (30), (31)]

Recognizing these early signs and symptoms is crucial for early detection and intervention, as it allows for the implementation of targeted strategies to support developmental progress and improve outcomes for individuals with ASD (32). By understanding the specific domains affected during early childhood, clinicians, caregivers, and educators can design interventions that address the unique challenges faced by children with ASD and promote their social, communicative, cognitive, and motor development.

2. Etiology and Risk Factors

Autism Spectrum Disorder (ASD) is a complex disorder that arises from the interplay of genetic and environmental factors (33). The genetic component of ASD is significant, with the disorder being highly heritable and involving a remarkable genetic heterogeneity (34). Over 800 ASD predisposition genes have been identified thus far, encompassing various biological processes such as chromatin remodelling, gene transcription regulation, cell growth and proliferation, ubiquitination, and neuronal - specific processes like synaptic organization, dendritic morphology, and axonogenesis. [(34), (35)] While there are fewer common single - gene mutations and chromosomal abnormalities associated with ASD, it is believed that multiple interacting genes of modest effect contribute to the risk. Genome - wide linkage analysis has identified several susceptibility loci and positional and functional candidate genes that potentially confer a risk for ASD. [(36)(37)(38)] Interestingly, many of these genes implicated in ASD also contribute to the genetic risk for other neurodevelopmental disorders like intellectual disability, schizophrenia, specific language impairment, epilepsy, and attention deficit hyperactivity disorder (ADHD). Understanding the specificity of these genes in relation to ASD remains a crucial challenge, necessitating further advances in research. (39)

Environmental factors also play a role in the pathogenesis of autism through their epigenetic effects. (40) These factors can be categorized into prenatal, natal, and postnatal risk factors. Prenatal risk factors(41) include advanced parental age, particularly paternal age, which has been identified as one of the most significant risk factors for autism [42](43)(44)(45)]. Maternal physical health conditions such as metabolic syndrome, bleeding, and maternal infections during pregnancy have also been associated with an increased risk of autism in children(46). Maternal mental health, parental behaviour, and communication patterns have shown associations with the formation of children’s personality and emotions, as well as the risk of mental disorders including autism(47). Maternal prenatal medication use has been linked to a 46% increased risk of autism in the fetus (48)(49)(50)(51). Studies examining the relationship between parental education and the risk of autism have yielded variable conclusions, with some confirming a correlation between lower levels of parental education and autism risk, while others indicate a stronger correlation between higher levels of parental education and the incidence of autism. (52)Natal risk factors include abnormal gestational age, with both preterm (<35 weeks) and post term pregnancy (>42 weeks) associated with a significantly increased risk of autism (53) (54) (55) (56). Fetal complications such as fetal distress and umbilical cord complications, including fetal nuchal cord and cesarean delivery, have been implicated in hypoxia (lack of oxygen) during birth, potentially increasing susceptibility to autism. In the postnatal period, low birth weight, jaundice, and postnatal infections, including meningitis, mumps, varicella, unknown fever, and ear infections within the first 30 days of...
life, have been correlated with a higher risk of autism. (57) (58) (59).

Understanding the interplay between genetic and environmental factors in the etiology of ASD is crucial for elucidating the underlying mechanisms and developing targeted interventions. Further research is needed to unravel the specific genetic contributions and interactions, as well as the precise impact of environmental factors on autism risk. Such knowledge can ultimately inform strategies for early detection, prevention, and personalized interventions for individuals with ASD.

**Diagnostic Assessment and Evaluation: Key Considerations in Autism Spectrum Disorders**

Pediatricians play a pivotal role in the early recognition of autism spectrum disorders (ASDs) as they often serve as the primary point of contact for parents seeking medical guidance. (60)(61) With increased awareness of the early signs of ASD due to widespread media coverage, parents are more likely to express concerns to their child’s pediatrician if they observe any published indicators. Over the past two decades, professionals specializing in autism have emerged, introducing the term autism spectrum disorders to encompass the diverse range of clinical characteristics that define this condition (60) (62).

Neuropathology and neuroimaging studies have contributed to our understanding of ASD by highlighting fundamental differences in brain growth and organization in individuals with ASD. (63)(64)(65) These differences are believed to originate in the prenatal period and persist throughout early childhood into adulthood. (66)(67) Notably, research has shown that children with ASD often exhibit increased head circumference and brain volume(68)(69)(70), with one study reporting larger - than - normal brain volumes in 90% of toddlers with ASD(71)(72)(73)(74)(75)(76) However, the challenge in recognizing ASD lies in the wide heterogeneity of features(78)(79)(80) observed among individual children.

When assessing for ASD, specific behavioural markers(81) can aid in early identification (82). These markers include a lack of appropriate gaze, absence of warm and joyful expressions in eye contact, a missing back - and - forth pattern of vocalizations between infant and parent typically observed around six months of age (where infants with ASD may continue vocalizing without regard for the parents speech) (81). Furthermore, children with ASD may display limited recognition of their mothers, fathers, or consistent caregiver’s voice, coupled with a disregard for vocalizations while maintaining a keen awareness of environmental sounds. Delayed onset of babbling past nine months of age, decreased or absent use of prespeech gestures (such as waving, pointing, and showing), and a lack of expressions like oh oh or & hush are additional signs to consider(83). Furthermore, children with ASD may exhibit a lack of interest or response to neutral statements. (82)

By recognizing these early behavioural indicators, healthcare professionals can facilitate early detection and timely interventions, leading to improved outcomes for children with ASD. Understanding the complex and diverse presentation of ASD is essential in guiding diagnostic assessments and evaluating the unique needs of each individual. Through comprehensive and multidisciplinary approach, we can enhance our diagnostic capabilities and provide targeted support to optimize the development and well - being of individuals with ASD.

**3. Conclusion**

In conclusion, this comprehensive review article provides a synthesized overview of existing research on autism, encompassing a wide range of topics related to the disorder. Through a thorough examination of various studies and publications, the review has explored crucial aspects such as the diagnostic process, etiological factors, prevalence rates, behavioral characteristics.

The findings from this review underscore the significance of early detection and intervention in improving outcomes for individuals with autism spectrum disorder (ASD). Early diagnosis enables the implementation of tailored interventions, leading to better social, communicative, and adaptive skills development. It highlights the need for increased awareness among healthcare professionals, educators, and parents to recognize early signs and ensure timely support.

Furthermore, the review emphasizes the multifactorial nature of autism, with genetic and environmental factors interacting to contribute to its complex etiology. While significant strides have been made in identifying specific genes associated with ASD, more research is required to fully comprehend the interplay between genetics and environmental influences in the development of the disorder.

**References**


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