



## A COMPREHENSIVE REVIEW OF NEURODEVELOPMENTAL DISORDER: AUTISM

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**ABSTRACT** Autism is diagnosed clinically based on the presence of core symptoms. However, caution is required when diagnosing autism because of non-specific manifestations in different age groups and individual abilities in intelligence and verbal domains. The earliest nonspecific signs recognized in infancy or toddlers include irritability, passivity, and difficulties with sleeping and eating, followed by delays in language and social engagement. Serotonin is the most commonly investigated neurotransmitter in Autism.

### KEYWORDS :

#### INTRODUCTION

Autism was first reported by Kanner (1943) with a clinical description of 11 children showing "extreme aloneness from the very starting of life, not responding to anything that comes to them from the outside world." He proposed the behavioral combination of autism, obsessiveness, stereotypy, and echolalia as childhood schizophrenia. However, until the 1980s, ASD was not accepted as an individual developmental disorder with a biological origin. Autism spectrum disorder (ASD) is a group of developmental disabilities characterized by abnormal social interaction and communication, and stereotyped behaviors with restricted interest.<sup>1</sup>

Autism is a clinically different neurodevelopmental disorder with growing occurrence. Recent statistics in the United States estimates that 1/68 children received the diagnosis of autism.<sup>2</sup>

#### DEFINITION

Autism disorder is identified by abnormal social communication with self-focus, impaired connectivity, organization and synaptogenesis.<sup>3</sup> Autism is defined as deferred societal communication skills and restricted, repetitive behaviors (RRB).<sup>4</sup>

Autism is a neurodevelopmental disorder with growing global health concern. This disorder is characterized by deficits in social and communication skills and restricted and repetitive behaviour; and these adversely impact quality of life of those affected as well as their families.<sup>5</sup>

#### EPIDEMIOLOGY

Globally, one in every 160 persons is estimated to live with Autism, contributing to 7.6 million disability life adjusted years.<sup>6</sup>

The current prevalence in the latest large-scale surveys is about 1%~2%. The prevalence has increased in the past two decades.<sup>7,8</sup>

#### DIAGNOSTIC CRITERIA

##### DSM 5 CRITERIA FOR DIAGNOSIS OF AUTISM<sup>9</sup>

<p><b>Social communication and interaction domain</b></p> <p><b>Deficits in social emotional reciprocity</b></p> <p>Abnormal social approach</p> <p>Failure of normal back and forth conversation Reduced sharing of interests, emotions, affect and response</p> <p>Total lack of initiation of social interaction</p> <p><b>2. Deficits in nonverbal communicative behaviors</b> Poorly integrated verbal and nonverbal communication</p> <p>Abnormalities in eye contact and body-language</p> <p>Deficits in understanding and use of nonverbal communication</p> <p>Total lack of facial expression or gestures</p> <p><b>3. Deficits in developing and maintaining relationships</b></p> <p>Difficulty making friends <b>or focus</b></p> <p>Apparent absence of interest in people Strong attachment to and/or preoccupation with unusual objects</p>	<p><b>Stereotyped or repetitive speech, motor movements, or use of objects</b></p> <p>Simple motor stereotypies</p> <p>Echolalia</p> <p>Repetitive use of objects</p> <p>Idiosyncratic phrases</p> <p><b>2. Excessive adherence to routines, ritualized patterns of behavior.</b></p> <p>Excessive resistance to change such as motoric rituals</p> <p>Insistence on same route or food</p> <p>Repetitive questioning or extreme distress at small changes</p> <p><b>3. Highly restricted, fixated interests that are abnormal in intensity</b></p> <p>Difficulties adjusting behavior to suit different situations Excessively circumscribed or pre-occupied interests</p> <p><b>4. Hyper- or hypo-reactivity to sensory input</b></p> <p>Unusual interest in sensory aspects of environment</p> <p>Apparent indifference to pain/heat/cold</p> <p>Adverse response to specific sounds or textures</p> <p>Excessive smelling or touching of objects Fascination with lights or spinning objects</p>
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#### ETIOLOGICAL FACTORS

Siblings born in families with an Autism subject have a 50 times greater risk of ASD, with a recurrence rate of 5%~8%.<sup>10</sup>

The concordance rate reaches up to 82%~92% in monozygotic twins, compared with 1%~10% in dizygotic twins. Genetic studies suggested that single gene mutations alter developmental pathways Of neuronal and axonal structures involved in synaptogenesis. Genetic causes including gene defects and chromosomal anomalies have been found in 10%~20% of individuals.<sup>11</sup>

Metabolic errors including phenylketonuria, creatine deficiency syndromes, adenylosuccinate lyase deficiency, and metabolic purine disorders are also account for less than 5% of individuals with ASD.<sup>12</sup> In recent years, some researchers suggest that Autism is the result of complex interactions between genetic and environmental risk

factors.<sup>13</sup>

#### CLINICAL FEATURES

Autistic children experiences mental retardation, emotional indifference, hyperactivity, aggression, self-injury, and repetitive behaviors such as body rocking or hand flapping. Repetitive, stereotyped behaviors are often accompanied by cognitive impairment, seizures or epilepsy, gastrointestinal complaints, disturbed sleep, and other problems. Differential diagnosis includes childhood schizophrenia, learning disability, and deafness.<sup>14</sup>

Autism is typically noticed in the first 3 years of life, with deficits in social behaviors and nonverbal interactions such as reduced eye contact, facial expression, and body gestures.<sup>15</sup>

At 12 months of age, individuals show atypical behaviors, across the

domains of visual attention, imitation, social responses, motor control, and reactivity.<sup>16</sup>

## DIAGNOSIS

The Autism Diagnostic Observation Schedule (ADOS) is a semistructured observational diagnostic interview that assesses the social communication skills and RRB of children suspected to have Autism.<sup>17</sup>

Autism is diagnosed clinically based on the presence of core symptoms. However, caution is required when diagnosing autism because of non-specific manifestations in different age groups and individual abilities in intelligence and verbal domains. The earliest nonspecific signs recognized in infancy or toddlers include irritability, passivity, and difficulties with sleeping and eating, followed by delays in language and social engagement. In the first year of age, infants later diagnosed with Autism cannot be easily distinguished from control infants. However, some authors report that about 50% of infants show behavioral abnormalities including extremes of temperament, poor eye contact, and lack of response to parental voices or interaction. At 12 months of age, individuals with Autism show atypical behaviors, across the domains of visual attention, imitation, social responses, motor control, and reactivity.<sup>18</sup>

## TREATMENT

Treatment of disabling symptoms such as aggression, agitation, hyperactivity, inattention, irritability, repetitive and self-injurious behavior may allow educational and behavioral interventions to proceed more effectively.<sup>19</sup>

TMS (Transcranial magnetic stimulation) lends itself to research for understanding, as well as potential treatment, of altered synaptic plasticity. TMS appears to be safe, but further research and clinical trials are Needed.<sup>20</sup>

Serotonin is the most commonly investigated neurotransmitter in ASD with reports of disruption of serotonergic innervation during development.<sup>21</sup>

## CONCLUSION

Autism is one of the neurodevelopmental disorder which is a result of complex interactions between genetic and environmental risk factors. characterized by deficits in social and communication skills and restricted and repetitive behaviour; and these adversely impact quality of life of those affected as well as their families.

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