

Review Articles

Autism Spectrum Disorder

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Abstract

Autism spectrum disorder (ASD) is the name for a group of developmental disorders. ASD affects social interaction, communication, interests and behavior. It includes a wide range, "a spectrum," of symptoms, skills, and levels of disability. It affects how a person acts and interacts with others, communicates, and learns. Children with ASD might have problems talking with others, or they might not look in the eye when one talks to them. They may often seem to be in their "own world." People with ASD often have these characteristics: ongoing social problems that include difficulty communicating and interacting with others; repetitive behaviors as well as limited interests or activities; symptoms that typically are recognized in the first two years of life; symptoms that hurt the individual's ability to function socially, at school or work, or other areas of life. Some people are mildly impaired by their symptoms, while others are severely disabled. According to the Centers for Disease Control and Prevention (CDC) around 1 in 68 children has been identified with some form of ASD. The symptoms are present before three years of age, although a diagnosis can sometimes be made after the age of three. More boys are diagnosed with the condition than girls. There is no "cure" for ASD, but speech and language therapy, occupational therapy, educational support, plus a number of other interventions are available to help children and parents.

Key words: Autism; Developmental disorders.

Introduction

Autism spectrum, also known as autism spectrum disorder (ASD), describes a range of conditions classified as neurodevelopmental disorders in the DSM-5, published in 2013. Individuals diagnosed with autism spectrum disorder must present two types

of symptoms: deficits in social communication and social interaction; restricted, repetitive patterns of behavior, interests or activities. Features of these disorders include: social deficits, communication difficulties, stereotyped or repetitive behaviors and interests, sensory issues, and in some cases, cognitive delays.

In 2015, a systematic review showed no association between caregiver interaction and language outcomes in ASD.³ Another controversial claim suggests that watching extensive amounts of television may cause autism. This hypothesis was largely based on research suggesting that the increasing rates of autism in the 1970s and 1980s were linked to the growth of cable television at this time.⁴

Epidemiology

It is estimated recently by the U.S. Center for Disease Control's that 1 out of every 68 children, or 14.7 per

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1,000, have some form of ASD as of 2010.⁵ Although prevalence rates vary for each of the developmental disorders in the spectrum. Autism prevalence has been estimated at 1-2 per 1,000. These rates are consistent across cultures and ethnic groups, as autism is considered a universal disorder.⁷

While rates of autism spectrum disorders are consistent across cultures, they vary greatly by gender, with boys affected far more frequently than girls. The average male-to-female ratio for ASDs is 4.2:1,⁸ affecting 1 in 70 males, but only 1 in 315 females.⁹ Females, however, are more likely to have associated cognitive impairment.¹⁰

Etiology

The exact cause of autism spectrum disorder (ASD) is currently unknown. It's a complex condition and may occur as a result of genetic predisposition (a natural tendency), environmental or unknown factors. Most researchers believe that certain genes a child inherits from their parents could make them more vulnerable to developing ASD. Cases of ASD have been known to run in families. No specific genes linked to ASD have been identified, but it may be a presenting feature of some rare genetic syndromes, including Fragile X syndrome, Williams syndrome and Angelman syndrome. It appears that there is no single gene that can account for autism. Instead, there seem to be multiple genes involved, each of which is a risk factor for components of the autism spectrum disorders.^{11,12,13} Despite extensive research, no reliable study has shown a link between autism spectrum disorder and any vaccines. In fact, the original study that ignited the debate years ago has been retracted due to poor design and questionable research methods.¹⁴

Several prenatal and perinatal complications have been reported as possible risk factors for autism. These risk factors include maternal gestational diabetes, maternal and paternal age over 30, bleeding after first trimester, use of prescription medication (e.g. valproate) during pregnancy, and meconium in the amniotic fluid.¹⁵ Low vitamin D levels in early development have been hypothesized as a risk factor for autism.¹⁶

Pathophysiology

Autism may involve a combination of brain enlargement in some areas and reduction in others¹⁷ that may be caused by abnormal neuronal growth and pruning during the early stages of prenatal and

postnatal brain development, leaving some areas of the brain with too many neurons and other areas with too few neurons.¹⁸ Some suggested an overall brain enlargement in autism, while others think in several areas of the brain, including the frontal lobe, the mirror neuron system, the limbic system, the temporal lobe, and the corpus callosum.¹⁹ Studies coincide with reports demonstrating abnormal patterns of cortical thickness and grey matter volume in those regions of autistic persons' brains.²⁰

Functions of the temporal lobe are related to many of the deficits observed in individuals with ASDs, such as receptive language, social cognition, joint attention, action observation, and empathy.²¹ It has also been suggested that ASD could be linked to mitochondrial disease (MD), a basic cellular abnormality with the potential to cause disturbances in a wide range of body systems.²²

It has been hypothesized that increased activity of serotonin in the developing brain may facilitate the onset of autism spectrum disorder.²³

Classification

A revision to autism spectrum disorder (ASD) was presented in the *Diagnostic and Statistical Manual of Mental Disorders* version 5 (DSM-5), released May 2013.²⁴ The new diagnosis encompasses previous diagnoses of autistic disorder, Asperger's disorder, childhood disintegrative disorder. Rather than categorizing these diagnoses, the DSM-5 has adopted a dimensional approach to diagnosing disorders that fall underneath the autism spectrum umbrella.²⁵

Autism forms the core of the autism spectrum disorders. Asperger syndrome is closest to autism in signs and likely causes;²⁶ unlike autism, people with Asperger syndrome have no significant delay in language development, according to the older DSM-4 criteria.²⁷

Clinical Features

Autism is characterized by persistent deficits in social communication and interaction across multiple contexts, as well as restricted, repetitive patterns of behavior, interests, or activities. These deficits are present in early childhood, and lead to clinically significant functional impairment. There is two main types of behaviors: "restricted / repetitive behaviors" and "social communication / interaction behaviors. Restrictive / repetitive behaviors may include:

repeating certain behaviors or having unusual behaviors; having overly focused interests, such as with moving objects or parts of objects; having a lasting, intense interest in certain topics, such as numbers, details, or facts. Social communication / interaction behaviors may include: getting upset by a slight change in a routine or being placed in a new or overly stimulating setting; making little or inconsistent eye contact; having a tendency to look at and listen to other people less often; rarely sharing enjoyment of objects or activities by pointing or showing things to others; responding in an unusual way when others show anger, distress, or affection; failing to, or being slow to, respond to someone calling their name or other verbal attempts to gain attention; repeating words or phrases that they hear, a behavior called *echolalia*; having facial expressions, movements, and gestures that do not match what is being said; having trouble understanding another person's point of view or being unable to predict or understand other people's actions.²⁸ There is also a unique form of autism called autistic savantism, where a child can display outstanding skills in music, art, and numbers with no practice.²⁹

Some of the language behaviors typically seen in children with autism may include repetitive or rigid language, specific interests in conversation, atypical language development, or poor nonverbal communication skills, including lack of eye contact and meaningful gestures and facial expressions. People with ASD may have other difficulties, such as being very sensitive to light, noise, clothing, or temperature. They may also experience sleep problems, digestion problems, and irritability.³⁰

Other characteristics of ASD include Restricted and Repetitive Behaviors (RRBs) which include a large range of specific gestures and acts; it can even include certain behavioral traits.³¹

Asperger syndrome was distinguished from autism in the DSM-IV by the lack of delay or deviance in early language development.³² Additionally, individuals diagnosed with Asperger syndrome did not have significant cognitive delays.³³

Complications and Comorbidities

Problems with social interactions, communication and behavior can lead to: problems in school and with successful learning; employment problems; inability to live independently; social isolation; stress

within the family; victimization and being bullied.³⁴ The most common medical condition occurring in individuals with autism spectrum disorders is seizure disorder or epilepsy, which occurs in 11-39% of individuals with ASD.³⁵ Tuberous sclerosis, a medical condition in which non-malignant tumors grow in the brain and on other vital organs, occurs in 1-4% of individuals with ASDs.³⁶

Intellectual disabilities are some of the most common comorbid disorders with ASDs. Recent estimates suggest that 40-69% of individuals with ASD have some degree of an intellectual disability.³⁷ Learning disabilities are also highly comorbid in individuals with an ASD. Approximately 25-75% of individuals with an ASD also have some degree of a learning disability.³⁸

Rates of comorbid depression in individuals with an ASD range from 4–58%.³⁹ Symptoms similar to those of attention deficit hyperactivity disorder (ADHD) can be part of an ASD diagnosis.⁴⁰ Sensory processing disorders is also comorbid with ASD, with comorbidity rates of 42–88%.⁴¹

Prognosis

Autism spectrum disorders are thought to follow two possible developmental courses. One course of development is more gradual in nature, in which parents report concerns in development over the first two years of life and diagnosis is made around 3–4 years of age. A second course of development is characterized by normal or near-normal development followed by loss of skills or regression in the first 2–3 years.⁴³

There continues to be a debate over the differential outcomes based on these two developmental courses. One group suggests that regression is associated with poorer outcomes while others report no differences between those with early gradual onset and those who experience a regression period.⁴⁴⁻⁴⁶ Some studies have shown that cognitive and language abilities at age 2.

Diagnosis

ASD could be diagnosed by looking at a child's behavior and development. Young children with ASD can usually be reliably diagnosed by age two. Older children and adolescents should be evaluated for ASD when a parent or teacher raises concerns based on watching the child socialize, communicate, and play. Diagnosing ASD in adults is not easy. In adults,

some ASD symptoms can overlap with symptoms of other mental health disorders, such as schizophrenia or attention deficit hyperactivity disorder (ADHD). Earlier screening might be needed if a child is at high risk for ASD or developmental problems. Those at high risk include children who: has a sister, brother, or other family member with ASD; have some ASD behaviors; were born premature, or early, and at a low birth weight. A reliable diagnosis can usually be made by the age of two.⁴⁸

Children who show some developmental problems during this screening process will be referred for another stage of evaluation. The diverse expressions of ASD symptoms pose diagnostic challenges to clinicians. Individuals with an ASD may present at various times of development (e.g., toddler, child, or adolescent), and symptom expression may vary over the course of development.⁴⁹ Furthermore, clinicians must differentiate among the different pervasive developmental disorders, and may also consider similar conditions, including intellectual disability not associated with a pervasive developmental disorder, specific language disorders, ADHD, anxiety, and psychotic disorders.⁵⁰

After a child shows initial evidence of ASD tendencies, psychologists administer various psychological assessment tools to assess for ASD.⁵¹ Among these measurements, the Autism Diagnostic Interview-Revised (ADI-R) and the Autism Diagnostic Observation Schedule (ADOS) are considered the “gold standards” for assessing autistic children. The ADI-R is a semi-structured parent interview that probes for symptoms of autism by evaluating a child’s current behavior and developmental history. The ADOS is a semistructured interactive evaluation of ASD symptoms that is used to measure social and communication abilities by eliciting several opportunities for spontaneous behaviors (e.g., eye contact) in standardized context. There isn’t a specific medical test to determine the disorder. Instead, a specialist may.⁵² In cases where a child with ASD also has a mental health problem, such as anxiety, a psychological treatment may be offered.

Management

The goal of treatment is to maximize child’s ability to function by reducing autism spectrum disorder symptoms and supporting development and learning. Early treatment for ASD and proper care can reduce individuals’ difficulties while helping them learn new

skills and make the most of their strengths. Early intervention during the preschool years can help child learn critical social, communication, functional and behavioral skills. However, a range of specialist educational and behavioral programmes can help children with ASD.⁵⁵ It can be difficult to know which intervention will work best for child, because each person with ASD is affected differently. Some types of intervention can involve hours of intensive work, and this isn’t always possible for many families because of the practical, emotional and financial commitments necessary.⁵⁶ In general, higher IQs are correlated with greater responsiveness to treatment and improved treatment outcomes. Children with autism spectrum disorder typically continue to learn and compensate for problems throughout life, but most will continue to require some level of support. Planning for child’s future opportunities, such as employment, college, living situation, independence and the services required for support can make this process.⁵⁷

The detailed assessment, management and co-ordination of care for children and young people with ASD should involve local specialist community-based multidisciplinary teams (sometimes called “local autism teams”) working together. The team may include: a pediatrician; mental health specialists, such as a psychologist and psychiatrist; a learning disability specialist speech and language therapist; an occupational therapist; education and social care services.⁵⁸ The parents of a child with ASD play a crucial role in supporting their child and improving their skills. If a child has ASD, it’s a good idea to find out as much as parents can about the condition. Although evidence-based interventions for autistic children vary in their methods, many adopt a psycho educational approach to enhancing cognitive, communication, and social skills while minimizing problem behaviors. It has been argued that no single treatment is best and treatment is typically tailored to the child’s needs.⁵⁹ Early treatment for ASD and proper care can reduce individuals’ difficulties while helping them learn new skills and make the most of their strengths.

Intensive, sustained special education programs and behavior therapy early in life can help children acquire self-care, social, and job skills. Available approaches include applied behavior analysis, developmental models, structured teaching, speech and language therapy, social skills therapy,

and occupational therapy.⁵⁹ Among these approaches, interventions either treat autistic features comprehensively, or focus treatment on a specific area of deficit.⁵⁸ Generally, when educating those with autism, specific tactics may be used to effectively relay information to these individuals. Utilizing as much social interaction as possible is key in targeting the inhibition autistic individuals experience concerning person-to-person contact.⁶⁰

The American Academy of Pediatrics (AAP) proposed new evidence-based recommendations for early interventions in ASD for children under 3. These recommendations emphasize early involvement with both developmental and behavioral methods, support by and for parents and caregivers, and a focus on both the core and associated symptoms of ASD.⁶¹

No medication can improve the core signs of autism spectrum disorder, but specific medications can help control symptoms. For example: sleeping problems – this may be treated with a medication such as melatonin; depression – this may be treated with a type of medication known as a selective serotonin reuptake inhibitor (SSRI); epilepsy – this may be treated with an anticonvulsant; attention – this may be treated with a medication such as methylphenidate; aggressive and challenging behavior, such as tantrums or self-harming – this may be treated with a type of medication called an antipsychotic if the behavior is severe or psychological treatments haven't helped. These medications can have significant side effects and are used only with caution by doctors specialized in that field. There's no evidence that special diets are an effective treatment for autism spectrum disorder. For growing children, restrictive diets can lead to nutritional deficiencies.⁶²

If a child's behavior is causing problems, they'll be assessed for possible triggers, such as a physical health condition, mental health problem, or environmental factors. In cases where a child with ASD also has a mental health problem, such as anxiety, a psychological treatment may be offered. Psychological treatments, such as cognitive behavioral therapy (CBT), involve talking to a trained therapist about thoughts and feelings, and discussing how these affect behavior and wellbeing.⁶³

Conclusions

Children with autism spectrum disorder typically continue to learn and compensate for problems

throughout life, but most will continue to require some level of support. Planning for child's future opportunities, such as employment, college, living situation, independence and the services required for support can make this process smoother.

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