What is Autism?

Autism, or autism spectrum disorder (ASD), refers to a broad range of conditions characterized by challenges with social skills, repetitive behaviors, speech and nonverbal communication. According to the Centers for Disease Control, autism affects an estimated 1 in 59 children in the United States today. We know that there is not one autism but many subtypes, most influenced by a combination of genetic and environmental factors. Because autism is a spectrum disorder, each person with autism has a distinct set of strengths and challenges. The ways, in which people with autism learn, think and problem-solve can range from highly skilled to severely challenged. Some people with ASD may require significant support in their daily lives, while others may need less support and, in some cases, live entirely independently. Several factors may influence the development of autism, and it is often accompanied by sensory sensitivities and medical issues such as gastrointestinal (GI) disorders, seizures or sleep disorders, as well as mental health challenges such as anxiety, depression and attention issues. Indicators of autism usually appear by age 2 or 3. Some associated development delays can appear even earlier, and often, it can be diagnosed as early as 18 months. Research shows that early intervention leads to positive outcomes later in life for people with autism.

What are the DSM-5 diagnostic criteria for autism?

In 2013, the American Psychiatric Association released the fifth edition of its Diagnostic and Statistical Manual of Mental Disorders (DSM-5). The DSM-5 is now the standard reference that healthcare providers use to diagnose mental and behavioral conditions, including autism.
Common signs of ASD?

Even as infants, children with ASD may seem different, especially when compared to other children their own age. They may become overly focused on certain objects, rarely make eye contact, and fail to engage in typical babbling with their parents. In other cases, children may develop normally until the second or even third year of life, but then start to withdraw and become indifferent to social engagement. The severity of ASD can vary greatly and is based on the degree to which social communication, insistence of sameness of activities and surroundings, and repetitive patterns of behavior affect the daily functioning of the individual. Social impairment and communication difficulties in many people with ASD find social interactions difficult. The mutual give-and-take nature of typical communication and interaction is often particularly challenging. Children with ASD may fail to respond to their names, avoid eye contact with other people, and only interact with others to achieve specific goals. Often children with ASD do not understand how to play or engage with other children and may prefer to be alone. People with ASD may find it difficult to understand other people’s feelings or talk about their own feelings. People with ASD may have very different verbal abilities ranging from no speech at all to speech that is fluent, but awkward and inappropriate. Some children with ASD may have delayed speech and language skills, may repeat phrases, and give unrelated answers to questions. In addition, people with ASD can have a hard time using and understanding non-verbal cues such as gestures, body language, or tone of voice. For example, young children with ASD might not understand what it means to wave goodbye. People with ASD may also speak in flat, robot-like or a sing-song voice about a narrow range of favorite topics, with little regard for the interests of the person to whom they are speaking. Many children with ASD engage in repetitive movements or unusual behaviors such as flapping their arms, rocking from side to side, or twirling. They may become preoccupied with parts of objects like the wheels on a toy truck. Children may also become obsessively interested in a particular topic such as airplanes or memorizing train schedules. Many people
with ASD seem to thrive so much on routine that changes to the daily patterns of life — like an unexpected stop on the way home from school — can be very challenging. Some children may even get angry or have emotional outbursts, especially when placed in a new or overly stimulating environment.

**What disorders are related to ASD?**

Certain known genetic disorders are associated with an increased risk for autism, including Fragile X syndrome (which causes intellectual disability) and tuberous sclerosis (which causes benign tumors to grow in the brain and other vital organs) — each of which results from a mutation in a single, but different, gene. Recently, researchers have discovered other genetic mutations in children diagnosed with autism, including some that have not yet been designated as named syndromes. While each of these disorders is rare, in aggregate, they may account for 20 percent or more of all autism cases. People with ASD also have a higher than average risk of having epilepsy. Children whose language skills regress early in life — before age 3 — appear to have a risk of developing epilepsy or seizure-like brain activity. About 20 to 30 percent of children with ASD develop epilepsy by the time they reach adulthood. Additionally, people with both ASD and intellectual disability have the greatest risk of developing seizure disorder.

**What causes ASD and what role do genes play?**

Scientists believe that both genetics and environment likely play a role in ASD. There is great concern that rates of autism have been increasing in recent decades without full explanation as to why. Researchers have identified a number of genes associated with the disorder. Imaging studies of people with ASD have found differences in the development of several regions of the brain. Studies suggest that ASD could be a result of disruptions in normal brain growth very early in development. These disruptions may be the result of defects in genes that control brain development and regulate how brain cells communicate with each other.
Autism is more common in children born prematurely. Environmental factors may also play a role in gene function and development, but no specific environmental causes have yet been identified. The theory that parental practices are responsible for ASD has long been disproved. Multiple studies have shown that vaccination to prevent childhood infectious diseases does not increase the risk of autism in the population. Twin and family studies strongly suggest that some people have a genetic predisposition to autism. Identical twin studies show that if one twin is affected, then the other will be affected between 36 to 95 percent of the time. There are a number of studies in progress to determine the specific genetic factors associated with the development of ASD. In families with one child with ASD, the risk of having a second child with the disorder also increases. Many of the genes found to be associated with autism are involved in the function of the chemical connections between brain neurons (synapses). Researchers are looking for clues about which genes contribute to increased susceptibility. In some cases, parents and other relatives of a child with ASD show mild impairments in social communication skills or engage in repetitive behaviors. Evidence also suggests that emotional disorders such as bipolar disorder and schizophrenia occur more frequently than average in the families of people with ASD. In addition to genetic variations that are inherited and are present in nearly all of a person’s cells, recent research has also shown that de novo, or spontaneous, gene mutations can influence the risk of developing autism spectrum disorder. De novo mutations are changes in sequences of deoxyribonucleic acid or DNA, the hereditary material in humans, which can occur spontaneously in a parent’s sperm or egg cell or during fertilization. The mutation then occurs in each cell as the fertilized egg divides. These mutations may affect single genes or they may be changes called copy number variations, in which stretches of DNA containing multiple genes are deleted or duplicated. Recent studies have shown that people with ASD tend to have more copy number de novo gene mutations than those without the disorder, suggesting that for some the risk of developing ASD is not the result of mutations in individual genes but rather spontaneous coding mutations.
across many genes. De novo mutations may explain genetic disorders in which an affected child has the
mutation in each cell but the parents do not and there is no family pattern to the disorder. Autism risk also
increases in children born to older parents. There is still much research to be done to determine the
potential role of environmental factors on spontaneous mutations and how that influences ASD risk.

Do symptoms for ASD change over time and how is it treated?

For many children, symptoms improve with age and behavioral treatment. During adolescence, some
children with ASD may become depressed or experience behavioral problems, and their treatment may
need some modification as they transition to adulthood. People with ASD usually continue to need services
and supports as they get older, but depending on severity of the disorder, people with ASD may be able to
work successfully and live independently or within a supportive environment. There is no cure for ASD.
Therapies and behavioral interventions are designed to remedy specific symptoms and can substantially
improve those symptoms. The ideal treatment plan coordinates therapies and interventions that meet the
specific needs of the individual. Most health care professionals agree that the earlier the intervention, the
better. Educational/behavioral interventions: Early behavioral/educational interventions have been very
successful in many children with ASD. In these interventions therapists use highly structured and intensive
skill-oriented training sessions to help children develop social and language skills, such as applied
behavioral analysis, which encourages positive behaviors and discourages negative ones. In addition,
family counseling for the parents and siblings of children with ASD often helps families cope with the
particular challenges of living with a child with ASD. Medications: While medication can’t cure ASD or
even treat its main symptoms, there are some that can help with related symptoms such as anxiety,
depression, and obsessive-compulsive disorder. Antipsychotic medications are used to treat severe
behavioral problems. Seizures can be treated with one or more anticonvulsant drugs. Medication used to
treat people with attention deficit disorder can be used effectively to help decrease impulsivity and hyperactivity in people with ASD. Parents, caregivers, and people with autism should use caution before adopting any unproven treatments. There are no medications that can cure ASD or treat the core symptoms. However, there are medications that can help some people with ASD function better. For example, medication might help manage high energy levels, inability to focus, depression, or seizures.

Medications might not affect all children in the same way. It is important to work with a health care professional that has experience in treating children with ASD. Parents and health care professionals must closely monitor a child’s progress and reactions while he or she is taking a medication to be sure that any negative side effects of the treatment do not outweigh the benefits. It is also important to remember that children with ASD can get sick or injured just like children without ASD. Regular medical and dental exams should be part of a child’s treatment plan. Often it is hard to tell if a child’s behavior is related to the ASD or is caused by a separate health condition. For instance, head banging could be a symptom of the ASD, or it could be a sign that the child is having headaches. In those cases, a thorough physical exam is needed. Monitoring healthy development means not only paying attention to symptoms related to ASD, but also to the child’s physical and mental health, as well. There are many different types of treatments available. For example, auditory training, discrete trial training, vitamin therapy, anti-yeast therapy, facilitated communication, music therapy, occupational therapy, physical therapy, and sensory integration.

According to reports by the American Academy of Pediatrics and the National Research Council, behavior and communication approaches that help children with ASD are those that provide structure, direction, and organization for the child in addition to family participation. A notable treatment approach for people with an ASD is called applied behavior analysis (ABA). ABA has become widely accepted among health care professionals and used in many schools and treatment clinics. ABA encourages positive behaviors and
discourages negative behaviors in order to improve a variety of skills. The child’s progress is tracked and measured. There are different types of ABA. Following are some examples: Discrete Trial Training (DTT) DTT is a style of teaching that uses a series of trials to teach each step of a desired behavior or response. Lessons are broken down into their simplest parts and positive reinforcement is used to reward correct answers and behaviors. Incorrect answers are ignored. Early Intensive Behavioral Intervention (EIBI) This is a type of ABA for very young children with an ASD, usually younger than five, and often younger than three. Pivotal Response Training (PRT) PRT aims to increase a child’s motivation to learn, monitor his own behavior, and initiate communication with others. Positive changes in these behaviors should have widespread effects on other behaviors. Verbal Behavior Intervention (VBI) VBI is a type of ABA that focuses on teaching verbal skills.

Dietary Approaches

Some dietary treatments have been developed by reliable therapists but many of these treatments do not have the scientific support needed for widespread recommendation. An unproven treatment might help one child, but may not help another. Many biomedical interventions call for changes in diet. Such changes include removing certain types of foods from a child’s diet and using vitamin or mineral supplements. Dietary treatments are based on the idea that food allergies or lack of vitamins and minerals cause symptoms of ASD. Some parents feel that dietary changes make a difference in how their child acts or feels. If you are thinking about changing your child’s diet, talk to the doctor first. Or talk with a nutritionist to be sure your child is getting important vitamins and minerals.

People with ASD often repeat behaviors and have narrow, obsessive interests. These types of behavior can affect eating habits and food choices, which can lead to the following health concerns. Limited food selection or strong food dislikes. Someone with autism may be sensitive to the taste, smell, color and
texture of foods. They may limit or totally avoid some foods and even whole food groups. Common dislikes include fruits, vegetables and slippery, soft foods. Not eating enough food. Kids with autism may have difficulty focusing on one task for an extended period of time. It may be hard for a child to sit down and eat a meal from start to finish. Constipation. This problem usually is caused by a child's limited food choices. It typically can be remedied through a high-fiber diet, plenty of fluids and regular physical activity. Medication interactions. Some stimulant medications used with autism, such as Ritalin, lower appetite. This can reduce the amount of food a child eats, which can affect growth. Other medications may increase appetite or affect the absorption of certain vitamins and minerals. If your child takes medication, ask your healthcare provider about possible side effects. Caring for a child with ASD can be challenging on many levels, and healthful eating is no exception. For children with ASD, a nutritious, balanced diet can make a world of difference in their ability to learn, how they manage their emotions and how they process information. Because children with ASD often have restricted diets as well as difficulty sitting through meal times, they may not be getting all the nutrients they need, particularly calcium and protein. You may have heard that a gluten- or casein-free diet can improve symptoms of ASD. Gluten is a type of protein found in wheat, rye and barley. Casein is a protein found in milk. Proponents of the diet believe people with autism have a "leaky gut," or intestine, which allows parts of gluten and casein to seep into the bloodstream and affect the brain and central nervous system. The belief is that this may lead to autism or magnify its symptoms. While some studies indicate that these diets may be effective for certain children, controlled scientific studies have not proven this to be true so more research is needed. Keep in mind that restrictive diets require careful planning to make sure your child's nutrition needs are being met. Consult with a registered dietitian nutritionist before making any drastic changes to your child's diet as there can be side effects and potential nutrient shortfalls when a gluten- or casein-free diet is self-prescribed.
References


2. https://www.autismspeaks.org/what-autism
