Autism Fact Sheet

What is autism spectrum disorder?

Autism spectrum disorder (ASD) refers to a group of complex neurodevelopment disorders characterized by repetitive and characteristic patterns of behavior and difficulties with social communication and interaction. The symptoms are present from early childhood and affect daily functioning.

The term “spectrum” refers to the wide range of symptoms, skills, and levels of disability in functioning that can occur in people with ASD. Some children and adults with ASD are fully able to perform all activities of daily living while others require substantial support to perform basic activities. The Diagnostic and Statistical Manual of Mental Disorders (DSM-5, published in 2013) includes Asperger syndrome, childhood disintegrative disorder, and pervasive developmental disorders not otherwise specified (PDD-NOS) as part of ASD rather than as separate disorders. A diagnosis of ASD includes an assessment of intellectual disability and language impairment.

ASD occurs in every racial and ethnic group, and across all socioeconomic levels. However, boys are significantly more likely to develop ASD than girls. The latest analysis from the Centers for Disease Control and Prevention estimates that 1 in 68 children has ASD.

What are some 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

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.

Repetitive and characteristic behaviors

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.

How is ASD diagnosed?

ASD symptoms can vary greatly from person to person depending on the severity of the disorder. Symptoms may even go unrecognized for young children who have mild ASD or less debilitating handicaps.

Autism spectrum disorder is diagnosed by clinicians based on symptoms, signs, and testing according to the Diagnostic and Statistical Manual of Mental Disorders-V, a guide created by the American Psychiatric Association used to diagnose mental disorders. Children should be screened for developmental delays during periodic checkups and specifically for autism at 18- and 24-month well-child visits.

Very early indicators that require evaluation by an expert include:

- no babbling or pointing by age 1
- no single words by age 16 months or two-word phrases by age 2
- no response to name
- loss of language or social skills previously acquired
- poor eye contact
- excessive lining up of toys or objects
- no smiling or social responsiveness

Later indicators include:

- impaired ability to make friends with peers
- impaired ability to initiate or sustain a conversation with others

- absence or impairment of imaginative and social play
- repetitive or unusual use of language
- abnormally intense or focused interest
- preoccupation with certain objects or subjects
- inflexible adherence to specific routines or rituals

If screening instruments indicate the possibility of ASD, a more comprehensive evaluation is usually indicated. A comprehensive evaluation requires a multidisciplinary team, including a psychologist, neurologist, psychiatrist, speech therapist, and other professionals who diagnose and treat children with ASD. The team members will conduct a thorough neurological assessment and in-depth cognitive and language testing. Because hearing problems can cause behaviors that could be mistaken for ASD, children with delayed speech development should also have their hearing tested.

What causes ASD?

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.

What role do genes play?

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 of autism change over time?

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.

How is autism treated?

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.

What research is being done?

The mission of the National Institute of Neurological Disorders and Stroke (NINDS) is to seek fundamental knowledge about the brain and nervous system and to use that knowledge to reduce the burden of neurological disease. The NINDS is a component of the National Institutes of Health (NIH), the leading supporter of biomedical research in the world. NINDS and several other NIH Institutes and Centers support research on autism spectrum disorder.

Nearly 20 years ago the NIH formed the Autism Coordinating Committee (NIH/ACC) to enhance the quality, pace, and coordination of efforts at the NIH to find a cure for autism. The NIH/ACC has been instrumental in promoting research to understand and advance ASD. The NIH/ACC also participates in the broader Federal Interagency Autism Coordinating Committee (IACC), composed of representatives from various U.S. Department of Health and Human Services agencies, the Department of Education, and other governmental organizations, as well as public members, including individuals with ASD and representatives of patient advocacy organizations. One responsibility of the IACC is to develop a strategic plan for ASD research, which guides research programs supported by NIH and other participating organizations.

NINDS and several other NIH institutes support autism research through the Autism Centers of Excellence (ACE), a trans-NIH initiative that supports large-scale multidisciplinary studies on ASD, with the goal of determining the causes of autism and finding new treatments. NINDS currently supports an ACE network focused on ASD and tuberous sclerosis complex (TSC). ASD occurs in approximately half of TSC patients. In particular, the ACE investigators are studying whether certain brain imaging and activity measures in infants diagnosed with TSC can predict the development of ASD. Such biomarkers could aid in understanding how and why ASD occurs in some children but not others, and help to identify patients who might benefit from early intervention. Other ACE centers and networks are investigating early brain development and functioning; genetic and non-genetic risk factors, including neurological, physical, behavioral, and environmental factors present in the prenatal period and early infancy; and potential therapies.

NINDS funds additional research aimed at better understanding the factors that lead to ASD, including other studies on genetic disorders associated with ASD, such as TSC, Fragile X Syndrome, Phelan-McDermid syndrome (which features such autism-like symptoms as intellectual disability, developmental delays, and problems with developing functional language), and Rett syndrome (a disorder that almost exclusively affects girls and is characterized by slowing development, intellectual disability, and loss of functional use of the hands). Many of these studies use animal models to determine how specific known mutations affect cellular and developmental processes in the brain, yielding insights relevant to understanding ASD due to other causes and discovering new targets for treatments.

NINDS researchers are studying aspects of brain function and development that are altered in people with ASD. For example, NINDS-funded researchers are investigating the formation and function of neuronal synapses, the sites of communication between neurons, which may not properly operate in ASD and neurodevelopmental disorders. Other studies use brain imaging in people with and without ASD to identify differences in brain connectivity and activity patterns associated with features of ASD. Researchers hope that understanding these alterations can help identify new opportunities for therapeutic interventions. Additional NINDS researchers are studying the relationship between epilepsy and autism.

Through the National Center for Advancing Translational Sciences (NCATS) Rare Disease Clinical Research Network (RDCRN), NINDS and other NIH Institutes and Centers support a research consortium focused on three rare genetic syndromes associated with ASD and intellectual disability, including TSC and syndromes involving mutations in the genes*SHANK3* (Phelan-McDermid syndrome) and *PTEN*. The goals of the consortium are to understand shared mechanisms across these syndromes, which may suggest common approaches to their treatment.

NINDS supports autism spectrum disorder research through clinical trials at medical centers across the United States to better our knowledge about ASD treatment and care. Information about participating in clinical studies can be found at the “NIH Clinical Trials and You” website at www.nih.gov/health/clinicaltrials. Additional studies can be found at www.clinicaltrials.gov. People should talk to their doctor before enrolling in a clinical trial.

More information about research on ASD supported by NINDS and other NIH Institutes and Centers can be found using NIH RePORTER (projectreporter.nih.gov), a searchable database of current and past research projects supported by NIH and other federal agencies. RePORTER also includes links to publications and resources from these projects.

Where can I get more information?

For more information on neurological disorders or research programs funded by the National Institute of Neurological Disorders and Stroke, contact the Institute's Brain Resources and Information Network (BRAIN) at:

BRAIN
P.O. Box 5801
Bethesda, MD 20824
800-352-9424

Information also is available from the following organizations:

Centers for Disease Control and Prevention (CDC)
U.S. Department of Health and Human Services
1600 Clifton Road
Atlanta, GA 30333
800-311-3435; 404-639-3311; 404-639-3543

National Institute of Child Health and Human Development (NICHD)
National Institutes of Health, DHHS
31 Center Drive, Rm. 2A32 MSC 2425
Bethesda, MD 20892-2425
301-496-5133

National Institute on Deafness and Other Communication Disorders (NIDCD)
National Institutes of Health, DHHS
31 Center Drive, MSC 2320
Bethesda, MD 20892-2320
301-496-7243; 800-241-1044; 800-241-1055 (TTY)

National Institute of Environmental Health Sciences (NIEHS)
National Institutes of Health, DHHS
111 T.W. Alexander Drive
Research Triangle Park, NC 27709
919-541-3345

National Institute of Mental Health (NIMH)
National Institutes of Health, DHHS
6001 Executive Blvd. Rm. 8184, MSC 9663
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301-443-4513; 866-415-8051; 301-443-8431 (TTY)

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