NAMI Autism Spectrum Disorders Fact Sheet

What are Autism Spectrum Disorders?
Autism Spectrum Disorders (ASDs) are complex developmental disorders of brain function. Each can affect a child’s ability through signs of impaired social interaction, problems with verbal and nonverbal communication, and unusual or severely limited activities and interest. These symptoms typically appear during the first three years of life. There is no cure for ASDs, but with appropriate early intervention, a child may improve social development and reduce undesirable behaviors.

ASDs affect an estimated two to six per 1,000 children and strike males about four times as often as females. They do not discriminate against racial, ethnic, or social backgrounds. ASDs are “spectrum disorders” that affect individuals differently and to varying degrees. The ASDs are Autism (the defining disorder of the spectrum), Asperger Syndrome, Pervasive Developmental Disorder Not Otherwise Specified (PDD-NOS), Rett Syndrome, and Childhood Disintegrative Disorder (CDD). The most severe cases are marked by extremely repetitive, unusual, self-injurious, and aggressive behavior. This behavior may persist over time and prove very difficult to change, posing a tremendous challenge to those who must live with, treat, and teach these individuals. The mildest forms of autism resemble a personality disorder associated with a perceived learning disability.

What are common signs of an ASD?
Children diagnosed with an ASD do not embrace the typical patterns of child development. Some hints of future problems may be apparent from birth, while in most cases, signs become evident when a child’s communication and social skills lag further behind other children of the same age. Some parents report the change as being sudden, and that their children start to reject people, act strangely, and lose language and social skills they had previously acquired.

ASDs are defined by a definite set of behaviors that can range from very mild to severe. Children with ASDs may fail to respond to their name and often avoid eye contact. They also have difficulty interpreting tone of voice or facial expressions and do not respond to others’ emotions or watch other people’s faces for cues about appropriate behavior. Many children will engage in repetitive movements such as rocking and hair twirling, or in self-injurious behavior such as nail biting or head-banging. They tend to speak later than other children and may refer to themselves by name instead of “I” or “me.” Some speak in 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. In summary, children do not outgrow ASDs, but studies show that early diagnosis and intervention lead to significantly improved outcomes.

Signs to look for include:

- Lack of or delay in spoken language (does not babble, point, or make meaningful gestures by one year; does not speak one word by 16 months; does
not combine two words by two years; does not respond to name; or loses language or social skill)
- Repetitive use of language and/or motor mannerisms (e.g., hand-flapping, twirling objects)
- Little or no eye contact
- Lack of interest in peer relationships
- Lack of spontaneous or make-believe play
- Persistent fixation on parts of objects
- Does not smile

Symptoms of an ASD do not remain static over a lifetime. About a third of children with an ASD—especially those with severe cognitive impairment and motor deficits—will eventually develop epilepsy. In many children, symptoms of an ASD improve with intervention or as the children mature. Some eventually lead normal or near-normal lives. ASDs in adolescence could worsen behavior problems in some children as they may become depressed or increasingly unmanageable. Parents should be aware and ready to adjust treatment to fit their child’s changing needs.

How is an ASD diagnosed?
Although much about ASDs is not known, the consensus is: the earlier the diagnosis, the earlier interventions and treatment can begin. Evidence over the past decade or more indicate that intensive, early intervention in optimal educational settings for at least two years during the preschool years result in improved outcomes in most young children. Currently, no medical test exists to determine if a child has or will develop an ASD. Therefore, when evaluating a child, clinicians rely on behavioral characteristics to make a diagnosis. Some of the characteristic behaviors of ASDs might be apparent in the first few months of a child’s life, but most often they appear at any time during the early years. A clinical diagnosis would come from an observed problem in at least one of the areas of communication, socialization, or restricted behavior before the age of three.

Diagnosis can be difficult for doctors because ASDs vary widely in severity and symptoms, and may go unrecognized, especially in mildly affected individuals or in those with multiple disabilities. Another consideration is that many of the behaviors associated with autism are shared by other disorders. Therefore, various medical tests may be ordered to rule out or identify other possible causes. For this reason, researchers have developed several sets of diagnostic criteria for ASDs. They include:

- Absence or impairment of imaginative and social play
- Impaired ability to make friends with peers
- Impaired ability to initiate or sustain a conversation with others
- Stereotyped, repetitive, or unusual use of language
- Restricted patterns of interests that are abnormal in intensity or focus
Diagnosis requires a two-stage process. The first stage involves developmental screening during “well child” check-ups. Several screening instruments have been developed to quickly gather information about a child’s social and communicative development within medical settings. The second stage of diagnosis must be done by a multidisciplinary team composed of a psychologist, a neurologist, a psychiatrist, a speech therapist, or other professionals who diagnose children with ASDs.

**What are the causes of ASDs?**

There is no known direct cause of the disorders, which is one of the reasons why ASDs continue to remain elusive to doctors in the field. ASDs are complex disorders and have remained relatively inaccessible for study. It can be said with great certainty, however, that autism is not the direct cause of a psychological disturbance caused by detached or uncaring “refrigerator” mothers, as once suggested in the 1940s by Dr. Bruno Bettelheim. There is also significant evidence from large-scale studies that refute the proposed link between thimerosal, a mercury-based preservative used in the measles-mumps-rubella (MMR) vaccine and autism.

Researchers believe that it is probably a combination of genetic and environmental factors. Studies of people with ASDs have found abnormalities in several regions of the brain, including the cerebellum, amygdala, hippocampus, septum and mamillary bodies. Neurons in these regions appear smaller than normal and have stunted nerve fibers, which may interfere with nerve signaling. This suggests that autism results from a disruption of normal brain development early in fetal development. Other studies suggest that people with an ASD have abnormalities of serotonin or other signaling molecules in the brain. These findings, however, are preliminary and require further study.

In recent studies it has even been suggested that some people have a genetic predisposition to ASDs. Scientists estimate that families with one child living with an ASD run the risk of approximately 5 to 10 percent of having a second child with one of the disorders—greater than the risk for the general population. Research continues into clues about which genes contribute to this increased susceptibility. Parents and other relatives of an autistic person show mild social, communicative, or repetitive behaviors that allow them to function normally but appear linked to ASDs. There is evidence that those who do not have a history with an ASD have a 0.1 to 0.2 percent chance that the family will have a child with an ASD.

**How are ASDs treated?**

At present, there is no specific cure for ASDs. Therapies or interventions are designed to remedy specific symptoms in each individual. The best-studied therapies include educational/behavioral and medical interventions, but these remedies do not ensure substantial improvement. The lack of proven treatments prompts many parents to pursue their own research, often using “trial and error.” Parents should use caution before subscribing to any particular treatment. Counseling for the families of people with autism also may assist them in coping with the disorder.
Educational / Behavioral Interventions
Educational and behavioral approaches are often a core feature of the overall treatment plan for children with an ASD. These strategies emphasize highly structured and often intensive, skill-oriented training that is tailored to the individual child. Therapists work with children to help them develop social and language skills. Recent evidence suggests that early intervention has a good change of favorably influencing brain development. Applied Behavior Analysis (ABA) is the most well-known of the behavioral approaches.

Medication
Doctors may prescribe a variety of drugs to reduce self-injurious behavior or other troublesome symptoms of ASDs, as well as associated conditions such as epilepsy and attention disorders. Most of these drugs affect levels of serotonin or other signaling chemicals in the brain. The medications most often used in the treatment of ASDs can generally be placed in one of the following groups: antipsychotic drugs, antidepressants, and stimulants.