

# Autism

## Making a Difference Today

Autism is a brain disorder that impairs a person's ability to think, feel, communicate, and relate appropriately to the outside world. These behaviors can range from mild to severely disabling, but their impact on the lives of individuals with the disorder and on their families is almost always devastating. Frequently, people with autism also have other debilitating brain disorders, including attention deficit hyperactivity disorder, obsessive-compulsive disorder, epilepsy, and depression.

Autism affects approximately one out of every 250 to 166 babies in the United States (about as many as 24,000 cases annually), making the disorder one of the more common and serious neurological conditions of early childhood. According to the Centers for Disease Control and Prevention, about 1.5 million Americans currently have autism or a related autism spectrum disorder such as Asperger syndrome, Rett syndrome, childhood disintegrative disorder, or pervasive developmental disorder not otherwise specified. And this number appears to be rising rapidly. Using statistics from the U.S. Department of Education and other governmental agencies, the Autism Society of America estimates that autism cases are growing at an annual rate of 10 percent to 17 percent. Whether this increase is due to changes in the way autism is diagnosed or to an actual increase in prevalence is not clear.

What is clear is that autism exacts an enormous emotional and social toll on affected individuals and their families. The financial burden to the U.S. economy is also immense. According to the Autism Society of America, the total societal cost of caring for people with autism is approximately \$90 billion per year.

### Greater Understanding

Fortunately, research funded by the National Institutes of Health (NIH) and others is helping scientists unlock the neurological mysteries of autism, thus opening the door to more effective methods of diagnosing and treating the disorder. Through studies involving families and twins, scientists have uncovered evidence that some people may be genetically vulnerable to autism. Several genetic "hotspots" or chromosomal regions associated with the disorder have been located, but identifying specific autism genes has proven extremely difficult. One of the most promising leads has been on chromosome 15, where a gene has been found that codes for a brain chemical involved in the autistic symptoms of repetitive behavior.

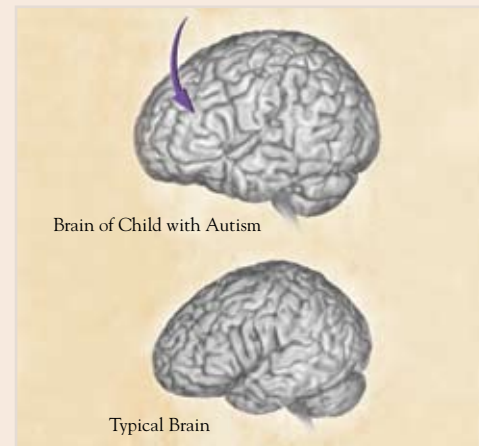
On another research front, scientists have used new brain imaging tools to make the discovery that children with autism tend to be born with brains that are average to slightly smaller than average. Before age two, however, these same children experience sudden, rapid and excessive brain growth, particularly in the cerebral cortex, where social, emotional, language, and other complex brain functions occur. The more abnormal the brain growth, the more severe the child's autistic symptoms.

Research into autism continues along a variety of other promising paths. Some scientists, for example, are exploring the role of serotonin and other brain chemicals in the development of autism. Others are investigating possible environmental causes of the disorder. Still others are attempting to identify precisely which regions of the brain are involved in various aspects of autism. Several brain structures have already been implicated. Research suggests that autism may result from the failure of various parts of the brain to work together.

### Diagnostic and Treatment Advances

Although there is no cure for autism, studies show that early diagnosis and intervention (before the age of three) results in better outcomes for children. The last decade has seen major advances in the standardization of diagnostic tools for autism and in the refinement and expansion of applied behavioral analysis, a one-on-one teaching approach that relies on reinforced practice of various skills and desirable behaviors.

Researchers also continue to explore possible drug therapies for autistic symptoms. A recent study funded by NIH found that the anti-psychotic medication risperidone not only decreased aggression among autistic children with severe behavioral problems, but also reduced their repetitive behaviors and increased their ability to socially interact.



The figure above shows that the brain of a 3-year-old child with autism is larger than that of a typical child. While the abnormal enlargement in brain size may not be this great in all autistic children, even slight abnormal brain overgrowth may reflect problems in the development of brain circuitry and brain function. Possibly newly discovered biological indicators of autism like brain overgrowth could be used together with behavioral signs to help confirm an autism diagnosis early. Currently, researchers are testing the use of measurements of brain size and other biomarkers to assist in diagnosing autism in the first year of life.

### Continued funding for research could lead to:

- Greater knowledge of the genetic and environmental factors that play a role in the development of autism.
- A clearer understanding of the function of various brain structures and chemicals in specific behaviors associated with autism.
- The discovery of a diagnostic marker—a biochemical abnormality that all people with autism might share—that could be detected by a simple chemical test or other screening technique in infants, before symptoms develop.
- More effective behavioral intervention programs and medications to help control the disabling symptoms of autism.

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## Making a Difference Tomorrow

Scientists are making progress in understanding the causes of autism and in developing better ways of diagnosing and treating it. A cure remains elusive, however, and the condition continues to devastate families.

Did you know that:

- Autism affects up to 1 in 166 babies born in the United States. Children in all racial, ethnic, and social groups are at risk.
- Autism usually becomes apparent by the age of three. Sometimes, the child seems “different” from birth; in other cases, the disorder appears in children who seemed to have been developing normally.
- Autism is 3 to 4 times more common in boys. Girls with the disorder, however, tend to have more severe symptoms.
- For reasons that are not clear, the number of American children diagnosed with autism is increasing at a rate of 10 percent to 17 percent per year.

### Research Brings Hope for the Future

The National Institutes of Health (NIH) has played a leading role in helping scientists advance their knowledge about this complicated neurological disorder.

With NIH funding, a library of DNA samples and clinical data from families with autism have been made available to molecular geneticists across the country. Armed with this data and new, cutting-edge technology, such as DNA microarray testing, scientists hope to discover the genes that lead to autism—and thus to develop better methods of diagnosing and treating the disorder.

Scientists also hope to better pinpoint the structure and function of brain regions involved in specific behaviors associated with autism, such as poor language skills and repetitive actions. More studies are also needed into what drives the excessive brain growth that occurs in many autistic children during the first two years of life. It is during this time that autistic behaviors first appear. Such research may enable doctors—for the first time—to identify infants and toddlers at risk for autism.

Diagnosing at-risk children early—before serious symptoms have developed and when therapeutic interventions might be most effective—is also the goal behind research into early biological biomarkers for autism. Recently, scientists found that children with autism had elevated levels of certain cells of the immune system in their blood—findings that add to growing evidence that autism may be a disorder of the immune system as well as of the brain. Research is now needed to determine if these immune abnormalities might be a very early marker of susceptibility to autism.

The role of environmental factors, such as exposure to polychlorinated biphenyls (PCBs), in the development of autism also needs more investigation. Although currently no study provides definitive evidence of an association between autism and vaccines, researchers continue to study that possibility.

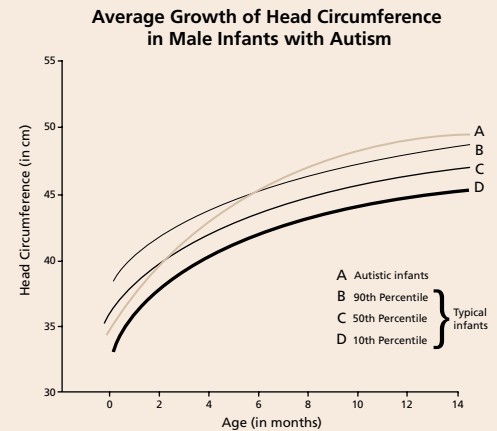
With NIH funding, researchers continue the valuable process of evaluating and improving the effectiveness of various behavioral screening tools and educational/behavioral intervention programs for autism. Research into better medications for treating severe behavioral symptoms of autism as well as related conditions, such as epilepsy and attention disorders, is also ongoing.

### Hope for Other Diseases

As scientists search for more effective methods of diagnosing and treating autism—and for a possible preventive cure—they also are learning more about a wide range of other neurological conditions that often exist concurrently with autism, including obsessive-compulsive disorder, depression, attention deficit hyperactivity disorder, epilepsy, and genetic disorders such as Fragile X. Research into autism thus benefits millions of Americans suffering from a variety of brain illnesses.

Autism is a highly complex neurological disorder. Only with continued funding will scientists be able to uncover new clues about how autism develops—clues that in the short term will greatly improve the lives of people with autism and their families and in the long term will lead to a cure.

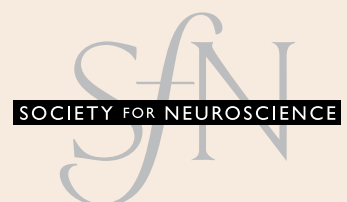
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Studies have indicated that autism is linked to two phases of head growth abnormality: a slightly reduced head circumference at birth and a sudden and excessive increase in head circumference in later development. This growth pattern may be one biological indication that, together with other behavioral signs, may lead to early detection of risk for autism.

### Already research has led to:

- The identification of several “hotspots” on the human chromosome associated with autism.
- A better understanding of which regions of the brain are involved in various aspects of autism.
- The discovery that the brains of autistic children tend to have a different growth pattern than those of non-autistic children.
- Better standardization of diagnostic tools.
- More effective behavioral intervention techniques.



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