

# N I C H D

National Institute  
of Child Health and  
Human Development

National Institutes of Health



## Autism Research at the NICHD



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## Autism Facts

The word “autism” has been in the news a lot lately. Congress held hearings on it; members of congress have formed their own caucus to focus on it. Television news programs and newspapers have done feature stories on it. Even popular television shows like *E/R* and *The West Wing* have had story lines about autism.

### But what *is* autism?

Researchers at the National Institute of Child Health and Human Development (NICHD), part of the National Institutes of Health (NIH), are trying to understand autism: what is it, what causes it, how to diagnose it, how to treat it. All these topics provide a focus for NICHD research. Autism is very complex. No two people with autism are exactly the same. No two people with autism respond to treatment in the same way. So research in autism is also very complex.

Some people have compared solving the puzzle of autism to peeling an onion: new insights reveal themselves one layer at a time.

Knowledge of autism is always changing, as research peels away more and more layers of this perplexing disease. This document explains what NICHD researchers who study autism have found out so far in their attempts to understand autism.

### What is autism?

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Autism is a complex biological disorder that generally lasts throughout a person's life. It is called a *developmental disability* because it starts before age three, in the developmental period, and causes delays or problems with many different ways in which a person develops or grows.

[www.nichd.nih.gov](http://www.nichd.nih.gov)

In most cases, autism causes problems with:

- Communication, both verbal (spoken) and nonverbal (unspoken)
- Social interactions with other people, both physical (such as hugging or holding) and verbal (such as having a conversation)
- Routines or repetitive behaviors, like repeating words or actions over and over, obsessively following routines or schedules for their actions, or having very specific ways of arranging their belongings

The symptoms of the disorder cut off people with autism from the world around them. Children with autism may not want their mothers to hold them. Adults with autism may not look others in the eye. Some people with autism never learn how to talk. These behaviors not only make life difficult for people who have autism, but also make life hard for their families, their health care providers, their teachers, and anyone who comes in contact with them.

## Why do some people get autism?

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Autism is not a disease that you “get,” the same way you can get the flu. Instead, scientists think autism has its beginnings before a person is even born. No one knows the exact cause or causes of autism, but scientists have some theories.

Some of the researchers in the Network on the Neurobiology and Genetics of Autism: Collaborative Programs of Excellence in Autism (CPEA), a worldwide research network

co-sponsored by the NICHD and the National Institute on Deafness and Other Communication Disorders (NIDCD), are focusing their efforts on possible genetic causes of autism. In 2000, scientists in the CPEA Network released the results of two studies that found genes were involved in autism. Additional papers were published in 2001 by CPEA researchers and other NIH-funded scientists as part of an international consortium on genetics research. These results lead researchers to believe that some people could have an error in their genes that makes them more likely to develop autism. The CPEA Network and other NICHD-supported researchers are also looking into other factors that could be involved in autism, in addition to genetics, including neurological, infectious, metabolic, immunologic, and environmental.

## How many people have autism?

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Currently, the exact number of cases of autism is not known, but estimates range from one-in-500 cases, to one-in-1,000 cases of autism diagnosed in the U.S. every year.

Initial studies done in the 1960s pointed to four-to-five cases of autism in 10,000 people, which is why autism was once thought of as a rare condition. However, dramatic increases in autism disorders in the U.S. and throughout the world clearly show that autism is not rare.

Keep in mind that changes in how autism is diagnosed, changes in what is considered autism, and changes in how autism cases are reported could account for some of the increases in the number of cases reported.

## Who usually gets autism?

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Current figures show that autism occurs in all racial, ethnic, and social groups. These statistics also show that boys are three-to-four times more likely to be affected by autism than girls are. In addition, if a family has one child with autism, there is a 5-to-10 percent chance that the family will have another child with autism. In contrast, if a family does not have a child with autism, there is only a 0.1-to-0.2 percent chance that the family will have a child with autism.

## When do people usually show signs of autism?

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In most cases, the symptoms of autism are measurable by certain screening tools at 18 months of age. However, parents and experts in autism treatment can usually detect symptoms before this time. In general, a formal diagnosis of autism can be made when a child is two, but is usually made when a child is between two and three, when he or she has a noticeable delay in developing language skills.

Recent studies show that at least 20 percent of children with autism experienced a “regression,” as reported by their parents. This means that the children had a mostly normal development, but then had a loss of social or communication skills. To date, however, there is little information about this type of regression, such as the age it seems to start, how severe it is, and what, if anything, triggers it. NICHD researchers are looking into a variety of possible causes for both early onset and regressive autism.

## Is there a link between autism and vaccines?

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To date, there has been no conclusive, scientific evidence that any part of a vaccine or any combination of vaccines causes autism. There is also no proof that any material used to make or preserve vaccines plays a role in causing autism.

In 2000, the Institute of Medicine (IOM) at the National Academy of Sciences, at the request of the Centers for Disease Control and Prevention (CDC) and the NIH, began a review of all the evidence related to the measles/mumps/rubella (MMR) vaccine and autism. This independent panel looked at completed studies, ongoing studies, and published medical and scientific papers, and heard testimony from experts on vaccines, autism, and digestive disorders to determine whether or not there was a link between autism and the MMR vaccine. The IOM concluded that the evidence it reviewed does not support a link between autism and the MMR vaccine. This and other conclusions from the IOM review were released in April 2001. For a full copy of the IOM report, visit the IOM Web site at [www.iom.edu](http://www.iom.edu) and look under “Recent Reports.”

The American Academy of Pediatrics (AAP) recently held a conference on the MMR vaccine and autism. Parents, scientists, and practitioners presented information on this topic to a multidisciplinary panel of experts. Based on its review, the AAP also found that the available evidence does not support the theory that the MMR vaccine causes or contributes to autism or related disorders.

The AAP policy statement appeared in the May 2001 issue of the journal *Pediatrics*.

Because there is no proven data to suggest a link between autism and vaccines, the National Immunization Program at the CDC, along with the AAP and the American Academy of Family Physicians, suggest that parents follow the recommended childhood immunization schedule that is published every year.

The CPEA Network, funded by the NICHD and the NIDCD, with additional funding from the CDC, is working to study autism and its relation to the MMR vaccine. CPEA researchers will compare vaccination records of groups of people with autism, to those who do not have autism, to see if the onset of autism symptoms was associated with getting the MMR vaccine or other vaccines. Lab tests in this study will also look for any signs of persistent infections that could be related to the MMR vaccine.

More information about this and other studies related to vaccines and autism is available in the NICHD fact sheet titled *Autism Research at the NICHD—Autism and the MMR Vaccine*. This and other fact sheets on autism are available on the NICHD Web site at [www.nichd.nih.gov/autism](http://www.nichd.nih.gov/autism), or from the NICHD Clearinghouse at 1-800-370-2943.

## **Do all people with autism have the same symptoms?**

Autism is a complex disorder that affects people differently. Because people with autism have a lot of similarities *and* differences, doctors now think of autism as a “spectrum”

disorder; so rather than being just one condition, autism is a group of conditions with a range of similar features. Doctors use the term “autism spectrum disorder (ASD)” to describe people with mild symptoms, severe symptoms, or symptoms that fall anywhere in between.

## **What conditions are included in the autism spectrum disorder (ASD) category?**

Currently, ASD includes:

- Autistic disorder (sometimes called “classic” autism)
- Asperger syndrome
- Childhood disintegrative disorder (CDD)
- Rett syndrome
- Pervasive Developmental Disorder Not Otherwise Specified (PDDNOS) or atypical autism

Depending on his or her specific symptoms, a person with autism can be in any one of these categories.

In 1999, NICHD-supported researchers identified the gene responsible for Rett syndrome, one of the conditions included in the ASD category. Rett syndrome occurs only in girls and causes them to develop autism-like symptoms after seemingly normal development. This discovery could lead to improved detection, prevention, and treatment of Rett syndrome.

Advances in detecting, preventing, and treating Rett syndrome may shed light on ways to understand and treat ASDs, including those aspects of ASD that may involve regression.

## Are there any behaviors that signal a need for a doctor to evaluate a child for autism?

A doctor should *definitely and immediately* evaluate a child for autism if he or she:

- Does not babble or coo by 12 months of age
- Does not gesture (point, wave, grasp, etc.) by 12 months of age
- Does not say single words by 16 months of age
- Does not say two-word phrases on his or her own (rather than just repeating what someone says to him or her) by 24 months of age
- Has *any* loss of *any* language or social skill at *any* age

## Are there other things that might be signs of autism?

There are a number of things that parents, teachers, and others who care for children can look for to determine if a child needs to be evaluated for autism. The following “red flags” could be signs that a doctor should evaluate a child for autism or a related communication disorder.

### “Red Flags” include...

- The child does not respond to his/her name.
- The child cannot explain what he/she wants.
- Language skills or speech are delayed.
- The child doesn't follow directions.
- At times, the child seems to be deaf.
- The child seems to hear sometimes, but not others.
- The child doesn't point or wave bye-bye.
- The child used to say a few words or babble, but now he/she doesn't.
- The child throws intense or violent tantrums.
- The child has odd movement patterns.
- The child is hyperactive, uncooperative, or oppositional.
- The child doesn't know how to play with toys.
- The child doesn't smile when smiled at.
- The child has poor eye contact.
- The child gets “stuck” on things over and over and can't move on to other things.
- The child seems to prefer to play alone.
- The child gets things for him/herself only.
- The child is very independent for his/her age.
- The child does things “early” compared to other children.
- The child seems to be in his/her “own world.”
- The child seems to tune people out.
- The child is not interested in other children.
- The child walks on his/her toes.
- The child shows unusual attachments to toys, objects, or schedules (i.e., always holding a string or having to put socks on before pants).
- Child spends a lot of time lining things up or putting things in a certain order.

## When should a doctor evaluate a child for autism?

Doctors should do a “developmental screening” at every well-baby and well-child visit, through the preschool years. In this screening, the doctor asks questions related to normal development that allow him or her to measure a specific child’s development. These questions are often more specific versions of the “red flags” listed on the previous page, such as *Does the child cuddle like other children?* Or, *Does the child direct your attention by holding up objects for you to see?* The doctor will also ask if the child has any features that were listed earlier as definite signs for evaluation for autism.

If the doctor finds that a child either has definite signs of autism, or has a high number of red flags, he or she will send the child to a specialist in child development or another type of health care professional, so the child can be tested for autism. The specialist will rule out other disorders and use tests specific to autism. Then he or she will decide whether a formal diagnosis of autism, ASD, or another disorder is appropriate.

## Is there a cure for autism?

To date, there is no cure for autism. However, there are a number of treatments that can help people with autism and their families lead more normal lives.

Individualized, intense interventions that begin as early as possible give people with autism their best chance for progress. Doctors suggest starting these treatments before a child is two-and-a-half or three to get the best and most lasting results. In some cases, treatment can help people with autism function at normal or near-normal levels.

## What are the treatments for autism?

Many families of children and adults with autism are finding new hope from a variety of treatments for autism. The list below does not include all of the possible treatments for autism. If you have a question about treatment, you should talk to a health care professional who specializes in caring for people with autism. Some treatments include:

- **Individualized Education Programs (IEPs)** are one effective way to prevent problem behaviors typically related to autism. IEPs involve a variety of interventions, including some of those mentioned below, and are designed to help a child or adult with autism to overcome his or her specific problems. Children with autism seem to respond very well to IEPs that are properly designed and systematically put into practice.
- **Comprehensive Treatment Programs** encompass a number of different theories about treating autism. These programs range from specific methods of learning, to applied

behavior analysis, to reaching certain developmental goals. In general, children need to be in this type of program for 15-40 hours a week, for two years or more, to change their behaviors and prevent problems.

- **Applied Behavior Analysis (ABA)** generally focuses on reducing specific problem behaviors and teaching new skills. Recently, ABA programs have broadened their scope to include what to do before or between episodes of problem behaviors, in addition to what to do during or after these episodes. By showing children or adults with autism how to handle things like a change in schedule, furniture that has been moved, and meeting new people, ABA removes these situations as triggers for problem behaviors.
- **Positive Behavioral Interventions and Support (PBS)** is an approach that tries to increase positive behaviors, decrease problem behavior, and improve the child's or adult's lifestyle. The PBS method looks at the interactions between people with autism, their environment, their behavior, and their learning processes to develop the best lifestyle for them.
- **Medications** can also be effective in improving the behavior or abilities of a person with autism. In general, these medications are called "psychoactive" because the drugs affect the brain of a person with autism. Medication is often used to deal with a specific behavior, such as reducing self-injurious behavior, which may allow the person with autism to focus on other things, like learning.

Many people with autism have other, treatable conditions in addition to their autism. Sleep disorders, seizures, allergies, and digestive problems are common among those with autism, but these problems can often be treated with medication. Treatment for these conditions may not cure autism, but it can improve the quality-of-life for people who have autism and their families.

## What special services are available to a family with a child who has been diagnosed with autism?

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According to Public Law 105-17: Individuals with Disabilities Act-IDEA (1997), the child's primary care provider is required to refer the family to an early intervention service. In addition, children age three and older are entitled by law to a free and appropriate public education. In some states, the law extends these services to all diagnosed children from birth to age three.

These services vary by state, but include special education and related services or treatment programs. If the child is under age three, the family should consult the zero-to-three service system in their community. The local school district can provide services for a family if the child is three or older. In either case, the local school district, the state education agency, and the local or state health departments should provide referrals for the necessary services.

The current service systems in many states are struggling to adjust to the increasing number of children diagnosed with autism. In many cases, however, the existing systems can't provide the level of care that families of people with autism want for their child, teenager, or adult with autism. There are a number of parents' organizations, both national and local, that can provide information about education and treatment services and how to get these services for a child. For a listing of these organizations, go to <http://www.nlm.nih.gov/medlineplus/autism.html>, or check the local phone book.

## Where can I go for more information about autism?

The NICHD Clearinghouse provides information on autism and autism research, and on other topics related to the health of children, adults, and families. The information specialists at the NICHD Clearinghouse are available at:

**Mail:** PO Box 3006, Rockville, MD 20847

**Phone:** 1-800-370-2943

**Fax:** 301-984-1473

**Email:** [NICHDClearinghouse@mail.nih.gov](mailto:NICHDClearinghouse@mail.nih.gov)

The NICHD Autism Web Page, [www.nichd.nih.gov/autism](http://www.nichd.nih.gov/autism), offers information on NICHD autism research, including the CPEA Network, current grants and funding mechanisms, ongoing clinical trials, and the NIH Autism Coordinating Committee. With a variety of information on topics related to autism, the NICHD Autism Web site is a good place to start your search for information. You can also comment on the *Autism Research at the NICHD* fact sheets through the Autism Web site.

## Are there other autism information resources I can consult?

In addition, the National Library of Medicine (NLM) offers a service called MEDLINEplus, which acts as a gateway for you to access all the NIH resources about autism. Some of these resources include the NICHD Autism Web Page and Web sites of other Institutes that are researching different aspects of autism. MEDLINEplus also provides links to recent news articles about autism, as well as links to non-government organizations that have autism as their main focus. To access MEDLINEplus, go to [www.nlm.nih.gov/medlineplus](http://www.nlm.nih.gov/medlineplus), and do a search for "autism."

