N I C H D

National Institute of Child Health and Human Development

National Institutes of Health

Autism Research at the NICHD

U.S. Department of Health and Human Services
National Institutes of Health

Autism and Genes

The **National Institute of Child Health and Human Development (NICHD)**, part of the National Institutes of Health (NIH), within the U.S. Department of Health and Human Services, is one of many federal agencies doing research on autism, including its possible causes.

In 1997, the NICHD and the National Institute on Deafness and Other Communications Disorders (NIDCD) started the *Network on the Neurobiology and Genetics of Autism: Collaborative Programs of Excellence in Autism (CPEA).* Researchers in this Network work to understand which genes might be involved in autism and how genes play a role in the condition. Working with other scientists around the world, the CPEA researchers have already learned a great deal about autism and genes.

What are genes?

Genes are pieces of DNA, material that contains all the information needed to "build" a person. Genes are **hereditary**, meaning parents pass genes on to their children.

Check the Glossary on pages 9-11 to learn how to say the **bolded** words and what they mean.

Most genetic material is found in the **nucleus** of a cell, a storage area that keeps these materials together in one place. The nucleus stores genetic materials in packages called **chromosomes**. Most people have 46 chromosomes in most of their cells: 23 from their mother and 23 from their father. Each chromosome is made up of genes.

Genes contain the information your body uses to make **proteins**, the body's building blocks. Proteins make up the structure of your organs and tissues; they are also needed for the body's chemical functions and pathways. Each protein performs a specific job in the body's different types of cells, and the information for making at least one protein is contained in a single gene.

The pattern or sequence of your genes is like a blueprint that tells your body how to build its different parts. For example, your genes control how tall you are, what color your eyes and hair are, and other features of your body and mind. Changes, or **mutations** in the blueprint can change how the body or mind grows/develops.

What is autism?

Autism is a complex neurobiological disorder of development that lasts throughout a person's life. It is sometimes called a *developmental disability* because it usually starts before age three, in the developmental period, and because it causes delays or problems in many different skills that arise from infancy to adulthood.

The main signs and symptoms of autism involve[1] language, social behavior, and behaviors concerning objects and routines:

- Communication—both verbal (spoken) and non-verbal (unspoken, such as pointing, eye contact, or smiling)
- Social interactions—such as sharing emotions, understanding how others think and feel (sometimes called empathy), and holding a conversation, as well as the amount of time a person spends interacting with others
- Routines or repetitive behaviors—often called **stereotyped** behaviors, such as repeating words or actions over and over, obsessively following routines or schedules, playing with toys or objects in repetitive and sometimes inappropriate ways, or having very specific and inflexible ways of arranging items

People with autism might have problems talking with you, or they might not want to look you in the eye when you talk to them. They may have to line up their pencils before they can pay attention, or they may say the same sentence again and again to calm themselves down. They may flap their arms to tell you they are happy, or they might hurt themselves to tell you they are not. Some people with autism never learn how to talk.

These behaviors not only make life difficult for people who have autism, but also take a toll on their families, their health care providers, their teachers, and anyone who comes in contact with them.

Because different people with autism can have very different features or symptoms, health care providers think of autism as a "*spectrum*" disorder—a group of disorders with a range of similar features. Based on their specific strengths and weaknesses, people with autism spectrum disorders (ASDs) may have mild symptoms or more serious symptoms, but they all have an ASD. This fact sheet uses the terms "*ASD*" and "*autism*" to mean the same thing.

What causes autism?

Scientists don't know exactly what causes autism.

Much evidence supports the idea that genetic factors—that is, genes, their function, and their interactions—are one of the main underlying causes of ASDs. But, researchers aren't looking for just one gene. Current evidence suggests that as many as 12 or more genes on different chromosomes may be involved in autism to different degrees.

Some genes may place a person at greater risk for autism, called **susceptibility**. Other genes may cause specific symptoms or determine how severe those symptoms are. Or, genes with mutations might add to the symptoms of autism because the genes or gene products aren't working properly.

Research has also shown that environmental factors, such as viruses, may also play a role in autism.

While some researchers are examining genes and environmental factors, other researchers are looking at possible neurological, infectious, metabolic, and immunologic factors that may be involved in autism.

Because the disorder is so complex, and because no two people with autism are exactly alike, autism is probably the result of many causes.

Why study genes to learn about autism?

Past research links autism and genes. For example:

- *Studies of twins with autism*—Scientists have studied autism in both **identical twins**—who are genetically the same—and **fraternal twins**—who are genetically similar, but not the same. When identical twins have autism, both have autism more than 60 percent[1] of the time, depending on the criteria used. When fraternal twins have autism, both have autism between 0 percent[2] and 6 percent of the time. If genes were not involved in autism, the rate of autism would be the same for both types of twins.
- *Family studies of autism*—Studies of family histories show that the chances a brother or sister of someone who has autism will also have autism is between 2 percent and 8 percent[3], which is much higher than in the general population. Also, some of the autism-like symptoms, such as delays in language development, occur more often[4] in parents and adult brothers and sisters of people with autism than in families who have no members or relatives with ASDs. Because members of the same family are more likely to share genes, something about these genes' sequences appears to be related to autism.
- *Diagnosable disorders and autism*—In about 5 percent[5] of autism cases, another single-gene disorder, chromosome disorder, or developmental disorder is also present. This type of co-occurrence helps researchers who are trying to pinpoint the genes involved in autism. Similar disorders or conditions with similar symptoms may have similar genetic beginnings. In cases of one disorder commonly occurring with another, it could be that one is actually a risk factor for the other. This kind of information can provide clues to what actually happens in autism. For example, many people with ASDs also have **epilepsy**, a condition marked by **seizures**. If scientists can understand what happens in epilepsy, they may also find clues to what happens in autism.

Based on these and other findings, scientists have long felt that there was a likely link between genes and autism.

But, how symptoms of ASDs affect family members and the wide variety of symptoms in ASDs tell researchers that they aren't looking for just one gene. So, even when scientists find the genes involved in autism, their work will be just beginning. They will still have to uncover what roles the genes play in the condition.

How do researchers look for the genes involved in autism?

Scientists generally use a combination of methods to find **candidate genes**—genes likely to be involved in autism.

Screen the whole genome.

A **genome** is all the genetic material in a person's cells—their DNA, their genes, and their chromosomes. Usually, researchers screen the genome of a family or a set of families that has more than one member with an ASD, to look for common features and differences. They look for so-called links between those diagnosed with ASDs and the genes within these families. Using "marker" locations—genes whose position in the genome is known—researchers can narrow down the location of genes involved in ASD. If a gene involved with autism is close to a particular marker, scientists can identify this gene by mapping it in relation to the known markers.

Conduct cytogenetic studies.

In a **cytogenetic** study, researchers stain chromosomes with a dye and then look at them under a microscope. The dye creates light and dark bands that are unique to each chromosome. Researchers compare the resulting bands of two people with autism, of one person with autism and one relative, or of one person with autism and one person not affected by ASD. These comparisons can point out similarities and differences between regions on the chromosome, which researchers can then study further based on the traits of that specific region.

Examine linkage ratios.

Researchers use this approach to to find **hot spots**—areas on chromosomes that may contain genes involved in autism. Hot spots are like neighborhoods on the chromosome where the genes involved in autism might "reside." In many cases, genes in the same area of a chromosome are tightly connected to one another, and this connection is hard to break. If the connection is present in more people than you might expect by chance, the situation is called **linkage disequilibrium**. Linkage disequilibrium helps researchers narrow their genetic search to find the spot in the chromosomal neighborhood where the gene might be.

Evaluate genes based on their known functions.

In some cases, researchers already know what the normal function of a specific gene or genes is. If that specific function is abnormal or incomplete in autism, researchers can look at the genes controlling that function in a person with autism to see what is different or missing. Or, they can look at what kinds of medications are useful in correcting or controlling that function to reduce the symptoms of autism. They can then study the chemical pathways that these medications effect to see what step might be

changed or missing in autism. Once they've found the pathway or the change, they can look at the genes that control these features for more information about autism. This approach is known as *genetic association analysis.*

What have researchers found by studying genes and autism?

Researchers in the CPEA Network and their colleagues around the world have learned a lot about autism through genetic studies, but they still have a great deal to learn. To date, some of their findings include the following.

Chromosomes where important genes are likely to be found

Using genome-wide screens, scientists have identified a number of genes that might be involved in autism. Although some analyses suggest that as many as 12 genes[6] might be involved in ASDs, the strongest evidence[7,13] points to areas on:

- *Chromosome 2*—Scientists know[7,10,11,12] that areas of chromosome 2 are the neighborhoods for "**homeobox**" or HOX genes, the group of genes that control growth and development very early in life. You have 38 different HOX genes in your chromosomal neighborhoods, and each one directs the action of other genes in building your body and body systems. Expression of these HOX genes is critical to building the brain stem and the cerebellum, two areas of the brain where functions are disrupted in ASDs.

- *Chromosome 7*—Researchers have found[6,7,8,9] a very strong link between this chromosome and autism. Their investigations now focus on a region called *AUTS1*, which is very likely associated with autism. Most of the genome studies completed to date have found that *AUTS1* plays some role in autism. There is evidence that a region of chromosome 7 is also related to speech and language disorders. Because ASDs affect these functions, autism may involve this chromosome.

- *Chromosome 13*—In one study, 35 percent[7,9] of families tested showed linkage for chromosome 13. Researchers are now trying to **replicate** these findings with other populations of families affected by autism.

- *Chromosome 15*—Genome-wide screens and cytogenetic studies show that a part of this chromosome may play a role in autism. Genetic errors on this chromosome cause **Angelman syndrome** and **Prader-Willi syndrome**, both of which share behavioral symptoms with autism. Cytogenetic errors on chromosome 15 occur[9] in up to 4 percent of patients with autism.

- *Chromosome 16*—Genes found on this chromosome control a wide variety of functions[7] that, if disrupted, cause problems that are similar or related to symptoms of autism. For example, a genetic error on this chromosome causes **tuberous sclerosis**, a disorder that shares many symptoms with autism, including seizures. So, regions on this chromosome may be responsible for certain similar behavioral aspects of the two disorders.

- *Chromosome 17*—A recent study found the strongest evidence[13] of linkage on this chromosome among a set of more than 500 families whose male members were diagnosed with autism. Missing or disrupted genes on this chromosome can cause problems, such as **galactosemia**, a metabolic disorder that, if left untreated, can result in mental retardation. Chromosome 17 also contains the gene for the serotonin transporter, which allows nerve cells to collect **serotonin**. Serotonin is involved in emotions and helps nerve cells communicate. Problems with the serotonin transporter can cause **obsessive-compulsive disorder (OCD)**, which is marked by recurrent, unwanted thoughts (obsessions) and/or repetitive behaviors (compulsions).

- *The X chromosome*—Two disorders that share symptoms with autism—**Fragile X syndrome** and **Rett syndrome**—are typically caused by genes on the X chromosome, which suggests that genes on the X chromosome may also play a role in ASDs. People generally have 46 chromosomes in most of their cells—23 from their mother and 23 from their father. After fertilization, the two sets match up to form 23 pairs of chromosomes. The chromosomes in the 23rd pair are called the "*sex chromosomes*," X and Y. Their combination determines a person's sex—males usually have one X and one Y chromosome, and females usually have two X chromosomes. The fact that more males than females have autism supports[5,9] the idea that the disorder involves genes on the X chromosome. Females may be able to use their other X chromosome to function normally, while males, without such a "back up" show symptoms of the condition.

Potential candidate genes

By focusing their studies on hot spots, researchers have narrowed their search for candidate genes. They need to do more work to understand how many genes are involved, and how these genes interact with each other and with the environment to cause autism. Researchers do have some promising leads—more leads than can be mentioned in this fact sheet, but these are a few.

Researchers have found evidence that autism may involve the *HOXA1* gene. *HOXA1*, a homeobox gene, plays a critical role in the development of important brain structures, cranial nerves, the ear, and the skeleton of the head and neck. Researchers know that the *HOXA1* gene is active very early in life—between the 20th and 24th days after conception—and that any problem with the gene's function causes problems with the development of these structures. Such problems may contribute to the features of autism.

In one study[10], nearly 40 percent of the persons with autism carried a specific mutation in the *HOXA1* gene sequence—nearly twice as many as those who had the same change, but who did not have autism and were not related to anyone with autism. In addition, 33 percent of those who did not have autism but were related to someone with autism also had the mutation in their *HOXA1* gene. These findings mean that autism does not result from genetics alone, but that some other factors are also involved in causing the condition. If researchers can confirm an association between this mutation and ASDs, they may be able to detect the mutation as an early test for autism, allowing important interventions to start as early in life as possible.

Another study[11] found that increased head size in ASD patients was associated with a different mutation in the *HOXA1* gene. About 20 percent of persons with autism have large head size. It is one of the most consistently reported physical features of persons with autism. Now researchers want to know whether the mutation affects head size in persons with autism only, or if it affects head size in general, regardless of ASD status.

Several other genes have come forward as potential candidates, including:

- *The Reelin (RELN) gene on chromosome 7* — This gene plays a crucial role in the development of connections between cells of the nervous system. Researchers think that abnormal brain connectivity plays a role in autism, which makes Reelin a good candidate. In addition, persons with autism and their parents and siblings have lower levels of certain types of the Reelin protein, which may mean that gene is not functioning normally.

- *The HOXD1 gene*—This homeobox gene is critical to the formation of certain brain structures. This gene is involved in **Duane syndrome,** a disorder that causes eye-movement problems and sometimes occurs with autism. In one study[12] of persons with autism, nearly 94 percent of participants had mutations in the same regions of *HOXD1*, which could mean that the region contributes to ASDs.

- *Gamma-amino-butyric acid (GABA) pathway genes*—GABA compounds are **neurotransmitters**, which means they help parts of the nervous system communicate with each other. GABA receptor genes are involved in early development of parts of the nervous system and help with communication between these parts throughout life. A problem in the GABA pathway can cause some of the symptoms of ASDs. For instance, epilepsy may result, in part, from low levels of GABA compounds. Many persons with autism also have epilepsy and also show low levels of GABA. Current research focuses on genes that, when their structure or function is incorrect, cause autism-like problems in mice.

- *Serotonin transporter gene on chromosome 17*— The serotonin transporter allows nerve cells to collect serotonin so that they can communicate. Serotonin is a neurotransmitter involved in depression, alcoholism/problem drinking, OCD, and other disorders. Research shows that persons with autism have higher-than-normal levels of serotonin—ranging between 25 percent and 50 percent[9,13] higher than persons without autism. This higher serotonin level may result from problems with the serotonin transporter that arise from errors in the gene.

Body chemicals that may play a role in autism

The body makes many chemicals that help it function correctly. When these chemicals are missing or incorrect, the body may have problems functioning properly, which may result in symptoms of autism or other disorders. Researchers are now trying to uncover how body chemicals might be involved in autism, so they can learn how the genes that make these chemicals might also play a role. Researchers are also studying whether medications might regulate or control these chemicals to create normal chemical levels. Normalizing the chemicals in a person with ASDs might reduce symptoms.

As mentioned earlier, GABA may play a role in autism and definitely plays a role in epilepsy. Levels of different types of GABA compounds are abnormally low in persons with autism. Researchers believe that these low levels may contribute to autism. In studies of mice, disrupting the GABA pathway causes seizures, extreme reactions to touch and sound, and stereotyped actions—symptoms also common in autism. Research now focuses on whether medications used to treat these problems can also reduce some of the symptoms of autism.

Another brain chemical mentioned earlier—**serotonin**—is also out-of-balance in many persons with autism. High serotonin levels may explain why persons with autism have problems showing emotion and handling sensory information, such as sounds, touch, and smells. Researchers now focus on whether medications that regulate serotonin levels may improve behavior in persons with autism. They also examine the genes that make and regulate serotonin and its pathway components to see if they can find any changes or patterns.

What does the future hold for studies of genes and autism?

Scientists in the CPEA Network and their colleagues who study the genetic mechanisms of autism hope that these studies will reveal the main cause or causes of autism. Doctors could then test for the gene or genes to detect autism early in life so that intervention can begin when it is most effective. Or, researchers could develop drugs that change or regulate the gene or genes to help normalize body chemicals and body functions.

Researchers share their information and their methods to see if other researchers can replicate their findings. Having several scientists get the same results "confirms" that discovery. Once confirmed, a discovery becomes the stepping stone to other discoveries. To date, however, not all genetic studies have gotten the same results. Therefore, additional work is still important.

Scientists also look beyond genes to find factors that may play a role in autism, including things in the environment. Environmental features can affect how genes function, which may contribute to the symptoms of ASDs. By understanding genetic and environmental causes of autism, scientists may better understand how to treat it and maybe even how to prevent it. Doctors and scientists continue to study genes, the environment, and gene-environment interactions until they solve the mysteries of autism.

Glossary

The word…	Is pronounced…	And means…
Angelman syndrome	Ayn-JELL-mann sinn-DROM	A genetic disorder caused by abnormal function of the gene UBE3A, located within a small region on chromosome 15. Characteristics include: developmental delay, lack of speech or minimal use of words; movement or balance disorder, usually ataxia of gait and/or tremulous movement of limbs, and any combination of frequent laughter/smiling; apparent happy demeanor; easily excitable personality, often with hand flapping movements; hypermotoric behavior; short attention span.
Candidate gene	kan-di-DATE jeen	A gene, located in a chromosome region suspected of being involved in a disorder, whose protein product suggests that it could be the gene in question.
Chromosome	kro-mu-SOM	One of the "packages" of genes and other DNA in the nucleus of a cell. Humans have 23 chromosome pairs, 46 in all. Each parent contributes one chromosome to each pair, so children get half of their chromosomes from their mothers and half from their fathers.
Cytogenetic	sigh-TOW-jenn-eh-tik	Study of chromosomes using a specific method that involves staining a chromosome and examining it under a microscope.
Duane syndrome	DWAYNE sinn-DROM	An inherited disorder characterized by inability of one or both eyes to turn outward beyond the midline. In some cases there is also a deficit of inward motility of the eye (turning toward the nose).
Epilepsy	epp-ih-LEPP-see	A brain disorder in which clusters of nerve cells, or neurons, in the brain sometimes signal abnormally. In epilepsy, the normal pattern of neuronal activity becomes disturbed, causing strange sensations, emotions, and behavior or sometimes convulsions, muscle spasms, and loss of consciousness.
Fragile X syndrome	FRA-jell EKS sinn-DROM	The most common form of inherited mental retardation. A mutation in a single gene, the *FMR1* gene on the X chromosome, causes the disorder, which can be passed from one generation to the next. Symptoms occur because the mutated gene cannot produce enough of a protein that is needed by the body's cells, especially cells in the brain, to develop and function normally.
Fraternal twins	frah-TURN-ul twinns	Twins resulting from the fertilization of two separate eggs. Fraternal twins share about 50 percent of their genes, just like siblings who are born at different times.
Galactosemia	guh-lak-toe-SEE-mee-uh	A rare disorder in which the body cannot process the sugar galactose, a by-product of milk metabolism. Buildup of galactose "poisons" the body, causing liver, kidney, and eye damage, and even death (if untreated).

Glossary (Continued)

The word…	Is pronounced…	And means…
Gene	jeen	Pieces of DNA. They contain the information for making a specific protein.
Genome	JEE-nom	All the DNA contained in an organism or a cell; includes both the DNA and chromosomes within the nucleus and the DNA outside the nucleus.
Hereditary	ha-RED-ih-tarry	A gene or trait passed down from parent to offspring.
Homeobox genes	HOE-mee-oh-boks jeenz	Genes found in almost all animals that control how and where parts of the body develop. Active very early in life, acting like a movie director by telling other genes when to act and when to stop in building the body.
Hot spots	hot spotz	Areas on chromosomes where mutations, activity, or recombination occurs with unusually high frequency.
Identical twins	eye-DEN-tik-ul twinns	Twins formed from the splitting of the same fertilized egg, so they share 100 percent of their genetic material.
Linkage disequilibrium	LINK-aj DISS-ee-kwel-ih-bree-um	An association of genes and/or markers near each other on a chromosome that is more than would be expected by chance. Linked genes and markers tend to be inherited together.
Mutation	my-TAH-shun	A permanent structural change in DNA. In most cases, DNA changes either have no effect or cause harm, but some mutations improve an organism's survival.
Neurotransmitter	nyur-OH-tranz-mitt-er	A substance that transmits nerve impulses between nerve cells.
Nucleus	NOO-klee-us	The central cell structure that houses chromosomes.
Obsessive-compulsive disorder (OCD)	ahb-SESS-iv kum-PUL-shen DISS-or-dr	A disorder characterized by recurrent, unwanted thoughts (obsessions) and/or repetitive behaviors or an urgent need to perform “rituals” (compulsions).
Prader-Willi syndrome	PRAY-derr WILL-ee sinn-DROM	An uncommon inherited disorder characterized by mental retardation, decreased muscle tone, short stature, emotional liability, and an insatiable appetite that can lead to life-threatening obesity. Caused by a missing part on the paternally derived chromosome 15.
Protein	PRO-teen	A large molecule made up of one or more chains of amino acids. Proteins perform a wide variety of activities in the cell and in the body.
Replicate	repp-li-KATE	Describes a situation in which many studies that use the same methods and steps have gotten the same outcome, suggesting that a finding is likely to be true.
Rett syndrome	RETT sinn-DROM	Results mostly from mutations in the *MECP2* gene on the X chromosome and occurs almost exclusively in girls. After seemingly normal development, affected girls develop problems with language, learning, coordination, and other brain functions.

Glossary (Continued)

The word…	Is pronounced…	And means…
Seizure	SEE-jyur	A sudden attack, often one of convulsions, as in epilepsy. Seizures don't necessarily involve movement or thrashing; they can also make someone seem as though they are frozen, unmoving.
Serotonin	serr-oh-TOE-ninn	A neurotransmitter that is found especially in the brain, blood serum, and stomach lining of mammals.
Stereotyped	STARE-ee-oh-tipd	An action that is repeated without change.
Susceptibility	suss-ept-ih-BULL	The state of being predisposed to, sensitive to, or of lacking the ability to resist manifestations of something (such as a pathogen, familial disease, or a drug); a person who is susceptible is more likely to show symptoms of a disorder.
Tuberous sclerosis	TOOB-er-us sklar-OH-siss	A rare, multi-system genetic disease that causes non-cancerous tumors to grow in the brain and on other vital organs such as the kidneys, heart, eyes, lungs, and skin. It commonly affects the central nervous system and results in a combination of symptoms including seizures, developmental delay, behavioral problems, skin abnormalities, and kidney disease.

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How can I get involved with studies of autism?

If you are interested in taking part in one of the CPEA studies, or if you want more information about one of the CPEA sites, visit *http://www.nichd.nih.gov/autism/cpea.cfm*. You and your family are welcome to take part in many different studies, but you can only take part in one genetics study at a time.

To find out what studies related to autism are currently looking for participants, go to *http://www.nichd.nih.gov/autism/research.cfm* and choose the "Autism clinical trials currently recruiting patients" link.

You can also visit *http://www.clinicaltrials.gov* or call 1-800-411-1222 for more information on federally funded studies that are seeking participants.

Where can I go for more information about genes and autism?

For more information about the CPEA Network, genetic studies, or autism research, contact the NICHD. The NICHD supports and conducts research on topics related to the health of children, adults, families, and populations, including autism and developmental disabilities. The mission of the NICHD is to ensure that every person is born healthy and wanted, that women suffer no harmful effects from the reproductive process, and that all children have the chance to fulfill their potential for a healthy and productive life, free of disease or disability, and to ensure the health, productivity, independence, and well-being of all people through optimal rehabilitation. You can contact the NICHD through the **NICHD Information Resource Center** at:

Mail: P.O. Box 3006, Rockville, MD 20847
Phone: 1-800-370-2943 (TTY: 1-888-320-6942)
Fax: (301) 984-1473
E-mail: NICHDInformationResourceCenter@mail.nih.gov (Please use AUTISM in the subject line)
Internet: http://www.nichd.nih.gov/autism

The National Library of Medicine also provides information on ASDs at http://www.nlm.nih.gov/medlineplus/autism.html. The NIH Web site also has information about ASDs at http://health.nih.gov/result.asp/62.

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