

# Fragile X Syndrome

## FACT SHEET

### What is fragile X syndrome?

Fragile X syndrome (FXS) is the most common known cause of intellectual disability (formerly referred to as mental retardation) that can be inherited, that is passed from parent to child. It is estimated that FXS affects about 1 in 4,000 boys and 1 in 6,000 to 8,000 girls. Both boys and girls can have FXS, but girls usually are more mildly affected.

### What causes FXS?

The cause of FXS is genetic. FXS occurs when there is a change in a gene on the X chromosome called FMR1. The FMR1 gene makes a protein needed for normal brain development. In FXS, the FMR1 gene does not work properly. The protein is not made, and the brain does not develop as it should. The lack of this protein causes FXS. Other Fragile X-associated Disorders (FXDs) can be present in the extended family, even if not currently evident. Talk with a genetic counselor for more information.

### What are some signs of FXS?

#### Children with FXS might:

- Sit up, crawl, or walk later than other children
- Have trouble with learning and solving problems
- Learn to talk later, or have trouble speaking
- Become very anxious in crowds and new situations
- Be sensitive about someone touching them
- Bite or flap their hands
- Have trouble making eye contact
- Have a short attention span
- Be in constant motion and unable to sit still
- Have seizures

#### Some children with FXS have certain physical features such as:

- A large head
- A long face
- Prominent ears, chin, and forehead
- Flexible joints
- Flat feet
- Macroorchidism (enlarged testicles in males; more obvious after puberty)

*These physical features tend to become more noticeable as the child gets older.*

### What conditions are common among children with FXS?

Children with FXS might have learning disabilities, speech and language delays, and behavioral problems such as attention-deficit/hyperactivity disorder (ADHD) and anxiety. Some boys can develop aggressive behavior. Depression can also occur. Boys with FXS usually have a mild to severe intellectual disability. Many girls with FXS have normal intelligence. Others have some degree of intellectual disability, with or without learning disabilities. Autism spectrum disorders (ASDs) occur more often among children with FXS.

### What can I do if I think my child has FXS?

Talk with your child's doctor or nurse. If you or your doctor think there could be a problem, the doctor can order a blood test for FXS or refer you to a developmental specialist or geneticist, or both. Also, contact your local early intervention agency (for children younger than 3 years of age) or public school (for children 3 years of age or older) to find out if your child qualifies for intervention services. To find out whom to call in your area, contact the National Information Center for Children and Youth with Disabilities at [www.nichcy.org/states.htm](http://www.nichcy.org/states.htm) or call the Centers for Disease Control and Prevention (CDC) at 1-800-232-4636.

In addition, CDC has links to information for families at [www.cdc.gov/ncbddd/single\\_gene/fragilex.htm](http://www.cdc.gov/ncbddd/single_gene/fragilex.htm).

Additional resources include the National Fragile X Foundation ([www.fragilex.org](http://www.fragilex.org)) and the FRAXA Research Foundation ([www.FRAXA.org](http://www.FRAXA.org)). CDC also supports the efforts of the Fragile X Clinical & Research Consortium ([www.FXCRC.org](http://www.FXCRC.org)) which can be reached through the National Fragile X Foundation.

While there is no cure for fragile X syndrome, therapies and interventions can improve the lives of those affected and of their families. It is very important to begin these therapies and interventions as early as possible to help your child reach his or her full potential. Acting early can make a real difference!



1-800-CDC-INFO

[www.cdc.gov/actearly](http://www.cdc.gov/actearly)

**Learn the Signs. Act Early.**





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## Facts about Fragile X Syndrome

Fragile X syndrome (FXS) is a genetic disorder. A genetic disorder means that there are changes to the person's genes (#). FXS is caused by changes in the fragile X mental retardation 1 (FMR1) gene. The FMR1 gene usually makes a protein called fragile X mental retardation protein (FMRP). FMRP is needed for normal brain development. People who have FXS do not make this protein. People who have other fragile X-associated disorders have changes in their FMR1 gene but usually make some of the protein.

FXS affects both males and females. However, females often have milder symptoms than males. The exact number of people who have FXS is unknown, but it has been estimated that about 1 in 5,000 males are born with the disorder. (1) (#references)

Learn how FXS is inherited » (/ncbddd/fxs/inherited.html)



Watch a video about what causes fragile X syndrome [VIDEO - 3.20 MB] (/ncbddd/fxs/video/FragileX-4.html) (Note: If you have difficulty viewing the clip, please send us an email (<mailto:cdcinfo@cdc.gov?subject=CDC%20Feature%20-%20Learn%20More%20About%20Fragile%20X%20Syndrome%20inquiry>)) or download a transcript  (/ncbddd/fxs/video/Fragile X transcript.txt).

## Signs and Symptoms

Signs that a child might have FXS include:

- Developmental delays (not sitting, walking, or talking at the same time as other children the same age);
- Learning disabilities (trouble learning new skills); and
- Social and behavior problems (such as not making eye contact, anxiety, trouble paying attention, hand flapping, acting and speaking without thinking, and being very active).

Males who have FXS usually have some degree of intellectual disability ([/ncbddd/dd/ddmr.htm](http://ncbddd/dd/ddmr.htm)) that can range from mild to severe. Females with FXS can have normal intelligence or some degree of intellectual disability. Autism spectrum disorders (ASDs) ([/ncbddd/autism/index.htm](http://ncbddd/autism/index.htm)) also occur more frequently in people with FXS.





## Diagnosis

FXS can be diagnosed by testing a person's DNA from a blood test. A doctor or genetic counselor ([/ncbddd/pediatricgenetics/genetic\\_counseling.html](http://ncbddd/pediatricgenetics/genetic_counseling.html)) can order the test. Testing also can be done to find changes in the FMR1 gene that can lead to fragile X-associated disorders.

A diagnosis of FXS can be helpful to the family because it can provide a reason for a child's intellectual disabilities and behavior problems. This allows the family and other caregivers to learn more about the disorder and manage care so that the child can reach his or her full potential. However, the results of DNA tests can affect other family members and raise many issues. So, anyone who is thinking about FXS testing should consider having genetic counseling prior to getting tested.

Learn more about diagnosing FXS at the following websites:

- American College of Medical Genetics (ACMG) policy statement on fragile X syndrome (<http://genetics.faseb.org/genetics/acmg/pol-16.htm>)  (<http://www.cdc.gov/Other/disclaimer.html>)
- National Fragile X Foundation – genetic testing (<http://www.fragilex.org/fragile-x-associated-disorders/testing/>)  (<http://www.cdc.gov/Other/disclaimer.html>)


## Treatments


There is no cure for FXS. However, treatment services can help people learn important skills. Services can include therapy to learn to talk, walk, and interact with others. In addition, medicine can be used to help control some issues, such as behavior problems. To develop the best treatment plan, people with FXS, parents, and health care providers should work closely with one another, and with everyone involved in treatment and support—which may include teachers, childcare providers, coaches, therapists, and other family members. Taking advantage of all the resources available will help guide success.

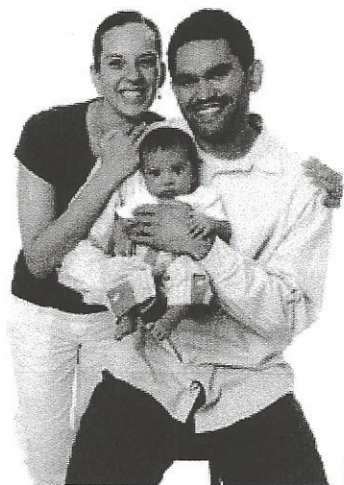


## Early Intervention Services

Early intervention services help children from birth to 3 years old (36 months) learn important skills. These services may improve a child's development. Even if the child has not been diagnosed with FXS, he or she may be eligible for services. These services are provided through an early intervention system in each state. Through this system, you can ask for an evaluation. In addition, treatment for particular symptoms, such as speech therapy for language delays, often does not need to wait for a formal diagnosis. While early intervention is extremely important, treatment services at any age can be helpful.

Learn more about treatments » (<http://www.nichd.nih.gov/publications/pubs/fragilex/sub13.cfm>)  
 (<http://www.cdc.gov/Other/disclaimer.html>)

Learn more about early intervention » (<http://nichcy.org/babies/overview>)   
(<http://www.cdc.gov/Other/disclaimer.html>)




## Finding Support

Having support and community resources can help increase confidence in managing FXS, enhance quality of life, and assist in meeting the needs of all family members. It might be helpful for parents of children with FXS to talk with one another. One parent might have learned how to address some of the same concerns another parent has. Often, parents of children with special needs can give advice about good resources for these children.

Remember that the choices of one family might not be best for another family, so it's important that parents understand all options and discuss them with their child's health care providers.

- Contact the National Fragile X Foundation at 1-800-688-8765 or [Treatment@FragileX.org](mailto:Treatment@FragileX.org) (<mailto:Treatment@FragileX.org>) to get information about treatments, educational strategies, therapies and intervention.
- Connect with a LINKS group (<http://www.fragilex.org/community/links-support-network/>)  (<http://www.cdc.gov/Other/disclaimer.html>) at the National Fragile X Foundation. LINKS groups are organized and run by parent volunteers and provide support to families.

- [Parent to Parent Programs \(http://www.p2pusa.org/\)](http://www.p2pusa.org/)  [provide information and emotional support to families of children who have special needs.](http://www.cdc.gov/Other/disclaimer.html)

## Related Pages

- [Intellectual Disability \(http://www.cdc.gov/ncbddd/dd/ddmr.htm\)](http://www.cdc.gov/ncbddd/dd/ddmr.htm)
- [Autism Spectrum Disorders \(http://www.cdc.gov/ncbddd/autism/index.html\)](http://www.cdc.gov/ncbddd/autism/index.html)
- [Attention-Deficit/Hyperactivity Disorder \(ADHD\) \(http://www.cdc.gov/ncbddd/adhd/\)](http://www.cdc.gov/ncbddd/adhd/)
- [Disability & Health \(http://www.cdc.gov/ncbddd/disabilityandhealth/index.html\)](http://www.cdc.gov/ncbddd/disabilityandhealth/index.html)
- [Pediatric Genetics \(http://www.cdc.gov/ncbddd/pediatricgenetics/\)](http://www.cdc.gov/ncbddd/pediatricgenetics/)
- [Genetic Counseling \(http://www.cdc.gov/ncbddd/pediatricgenetics/genetic\\_counseling.html\)](http://www.cdc.gov/ncbddd/pediatricgenetics/genetic_counseling.html)
- [CDC's National Center on Birth Defects and Developmental Disabilities \(http://www.cdc.gov/ncbddd\)](http://www.cdc.gov/ncbddd)

## References

1. Am J Hum Genet. 2009 Oct;85(4):503-14. Incidence of fragile X syndrome by newborn screening for methylated FMR1 DNA. Coffee B, Keith K, Albizua I, Malone T, Mowrey J, Sherman SL, Warren ST.

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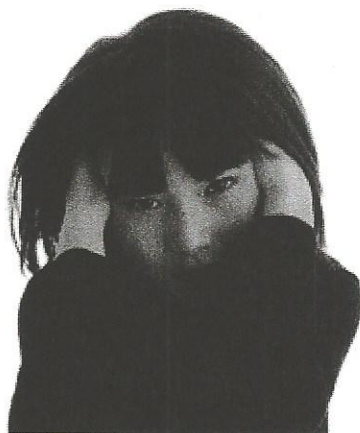
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## Related Concerns


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Fragile X Syndrome (FXS) often occurs with other conditions. Some of these conditions include anxiety, attention-deficit/hyperactivity disorder (ADHD), autism spectrum disorders, depression, difficult peer relationships, intellectual disabilities, and learning disabilities.

### Anxiety

There are many different types of anxiety disorders with many different causes and symptoms. These include generalized anxiety disorder, obsessive-compulsive disorder (OCD), panic disorder, post-traumatic stress disorder, separation anxiety, and different types of phobias. Separation anxiety is most common among young children. These children feel very worried when they are apart from their parents.

[Learn more about anxiety » \(http://www.nimh.nih.gov/health/publications/anxiety-disorders/index.shtml\)](http://www.nimh.nih.gov/health/publications/anxiety-disorders/index.shtml)   [\(http://www.cdc.gov/Other/disclaimer.html\)](http://www.cdc.gov/Other/disclaimer.html)

### Attention-Deficit/Hyperactivity Disorder (ADHD)

Children with attention-deficit/hyperactivity disorder (ADHD) have trouble paying attention and controlling impulsive behaviors. They might act without thinking about what the result will be and, in some cases, they are also overly active. It is normal for children to have trouble focusing and behaving at one time or another. However, these behaviors continue beyond early childhood (0-5 years of age) among children with ADHD. Symptoms of ADHD can continue and can cause difficulty at school, at home, or with friends.

[Learn more about ADHD » \(/ncbddd/adhd/index.html\)](/ncbddd/adhd/index.html)

### Autism Spectrum Disorders (ASDs)

Autism spectrum disorders (ASDs) are a group of developmental disabilities (</ncbddd/dd/>) that can cause significant social, communication and behavior challenges. People with ASDs handle information in their brain differently than other people.


ASDs are "spectrum disorders." That means ASDs affect each person in different ways, and can range from very mild to severe. People with ASDs share some similar symptoms, such as problems with social interaction. But there are differences in when the symptoms start, how severe they are, and the exact nature of the symptoms.

[Learn more about ASDs » \(http://www.cdc.gov/ncbddd/autism/index.html\)](http://www.cdc.gov/ncbddd/autism/index.html)

## Depression

Everyone feels worried, anxious, sad, or stressed from time to time. However, if these feelings do not go away and they interfere with daily life (for example, keeping a child home from school or other activities, or keeping an adult from working or attending social activities), a person might have depression. Having either a depressed mood or a loss of interest or pleasure for at least weeks might mean that someone has depression. Children and teens with depression might be irritable instead of sad. Depression can be treated with counseling and medication.

[Learn more about depression](#)

» (<http://www.nimh.nih.gov/health/publications/depression/index.shtml>) 

(<http://www.cdc.gov/Other/disclaimer.html>)

## Difficult Peer Relationships

FXS can have many effects on a child's development. It can make childhood friendships, or peer relationships, very difficult. These relationships contribute to children's immediate happiness and may be very important to their long-term development.

Children with FXS might have difficulty in their peer relationships, for example, being rejected by peers or not having close friends. In some cases, children with peer problems may also be at higher risk for anxiety, behavioral and mood disorders, substance abuse and delinquency as teenagers.

## Intellectual Disability

People with intellectual disability have a significantly below-average score on a test of mental ability or intelligence and limitations in the ability to function in areas of daily life, such as communication, self-care, and getting along in social situations and school activities.


Children with intellectual disability can and do learn new skills, but they develop more slowly than children with average intelligence and adaptive skills. There are different degrees of intellectual disability, ranging from mild to severe. A person's level of intellectual disability can be defined by their intelligence quotient (IQ), or by the types and amount of support they need.

[Learn more about intellectual disability » \(/ncbddd/dd/ddmr.htm\)](/ncbddd/dd/ddmr.htm)



## Learning Disabilities

There are many kinds of learning disabilities (also called learning disorders). They can range from mild to severe and affect each person in different ways. Learning disabilities may affect a person's ability to read, write, listen, talk, reason, do math, and pay attention.

Learn more about learning disabilities » (<http://nichcy.org/disability/specific/ld>)   
(<http://www.cdc.gov/Other/disclaimer.html>)

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- [Intellectual Disability](http://www.cdc.gov/ncbddd/dd/ddmr.htm) (<http://www.cdc.gov/ncbddd/dd/ddmr.htm>)
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- [CDC's National Center on Birth Defects and Developmental Disabilities](http://www.cdc.gov/ncbddd) (<http://www.cdc.gov/ncbddd>)

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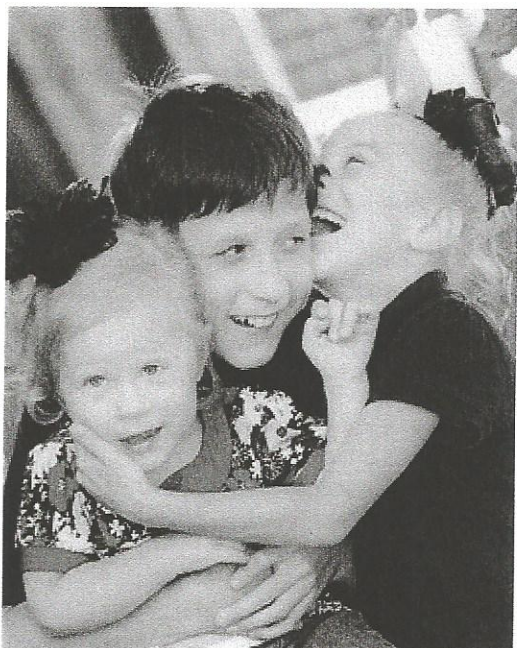
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## My Story

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### Real Stories from People living with Fragile X Syndrome

#### Rachael's Story



"We had a miracle last week. My youngest child Joslin grabbed my hand and pulled me to the refrigerator. My husband and I still had to guess what she wanted, but it's a big deal that she's pulling us to the refrigerator. Kids with fragile X just can't communicate what they want because of speech delays or hyperactivity. We have to rejoice in the little things.

"All three of our children have fragile X syndrome with the full mutation, and they all have Attention-Deficit/Hyperactivity Disorder. There's occupational therapy, physical therapy, and speech therapy at school.

"Brighton, our oldest, has grown out of a lot of his needs, but he still has some speech therapy. Avery, our middle child, has occupational therapy just for her sensory needs. Sometimes we take her down to a hammock we have hanging in our basement and do a lot of swinging, or we have a soft baby brush and we brush her arms and her legs if things are really bad. She needs that sensory input. For her, falling apart can be anything from rocking and bouncing and humming to total mayhem where she hurls herself into the wall or furniture, and throws things. Joslin, our youngest, has her therapies at an early intervention center.

"I wish we had the resources to have specialists come to our house but we don't. The burden falls on us as parents to fill those gaps, so that's added pressure. We tell our family what we're working on with the kids and they'll help Brighton with his speech, or if Avery starts falling apart they'll give her a good hug for the sensory input. Having that support system has made a world of difference for us.

"Fragile X is so hard because it's genetic. We had no idea it was in our family. When Brighton was diagnosed at five, we had Avery tested. She was nine months old. Marc and I didn't understand the genetic piece and when I learned I was the carrier, I felt like I wanted to die. My dad is a carrier and so are my sisters. So far none of them have children with fragile X, but there's always that chance. Since I have daughters, I also worry if they'll have the ovarian insufficiency that can come with fragile X, and if they'll be able to have children of their own.

"At the beginning when Brighton was diagnosed, I wish fragile X hadn't been such a foreign concept. I felt so alone when I got the diagnosis. I also wish that in my time of mourning I would have been able to look into the future and see how happy my kids are and how happy our lives are. There are tough times, there really are. But there's so much joy."

CDC would like to thank Rachel for sharing her personal story.

***If you would like to share your personal story, please contact us at [cdcinfo@cdc.gov](mailto:cdcinfo@cdc.gov) (<mailto:cdcinfo@cdc.gov>)***

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- [Intellectual Disability \(http://www.cdc.gov/ncbddd/dd/ddmr.htm\)](http://www.cdc.gov/ncbddd/dd/ddmr.htm)
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