

Fragile X Syndrome

Fragile X Syndrome (FXS) is a genetic condition that can cause learning, behavior, and developmental challenges, as well as various physical characteristics. It is the most common inherited cause of intellectual disability and is the leading known genetic cause of autism. FXS occurs when a change in the *FMR1* gene on the X chromosome affects how the brain develops and functions.

While there is no cure, many people with Fragile X lead full, meaningful lives with the right combination of therapies, school supports, and medical care. Families often discover strength, resilience, and community through their journey. Here we explain and guide you through what Fragile X syndrome is, how it's inherited, its signs and symptoms, and the treatments and resources that can help. We have several additional pages on the top topics within FXS.

Fragile X 101

A Guide for the Newly Diagnosed and Those Already Living with Fragile X

This guide helps answer common physical, cognitive, and behavioral questions. You'll also learn:

— How Fragile X is inherited.

— Available treatment options.

— How females are affected vs. males.

— About the support you can receive from the NFXF.

Fragile X 101

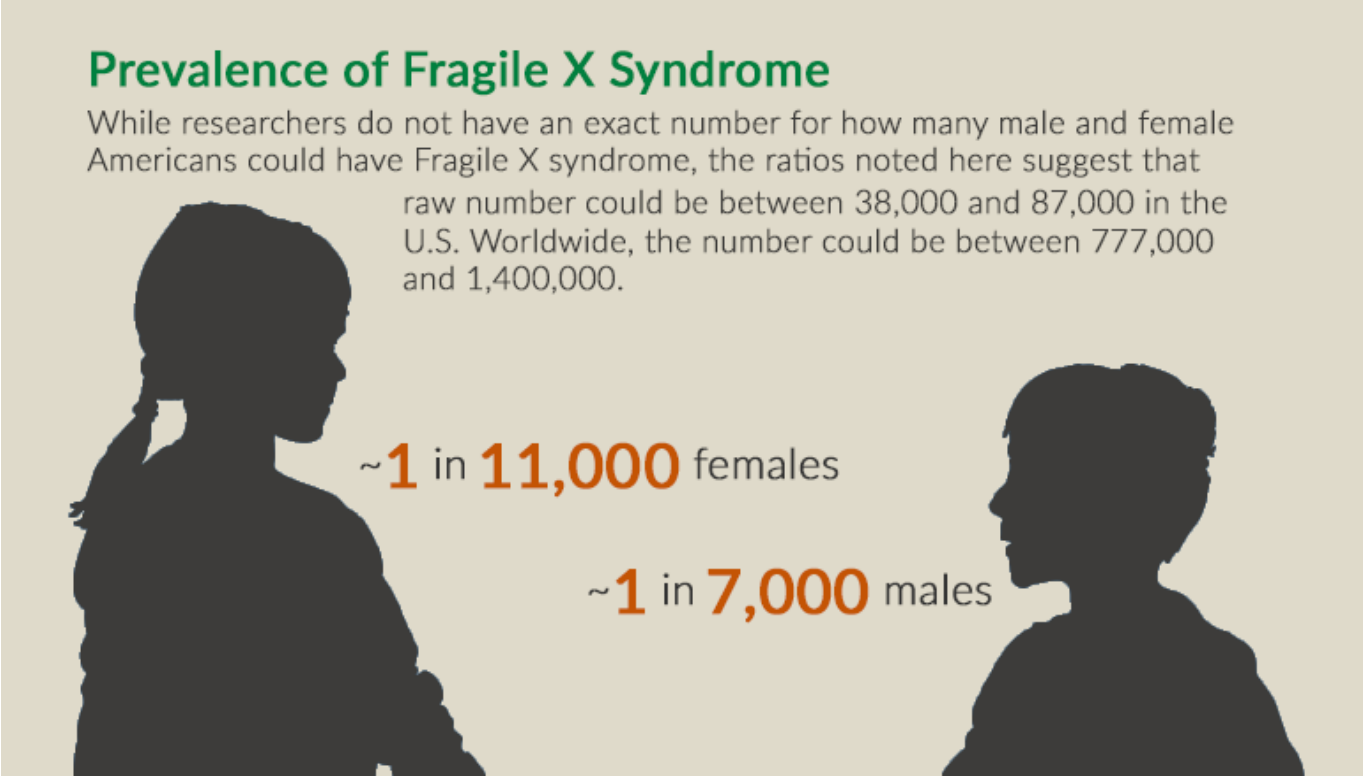
Females with the Fragile X premutation have a 50% chance of passing the mutation to each of their children, while males with the Fragile X premutation will pass it to all of their daughters (and none of their sons).

Fragile X syndrome occurs in both genders. FXS is more common in males than females, and males can experience more severe symptoms than females, though that is not always true.

Exactly why males are more often and more severely affected is unknown, but it's believed to be because females have two X chromosomes and males have only one — can an impacted *FMR1* gene on one X chromosome be compensated for by a typical *FMR1* gene on the other? Because males have only one X, if they have an impacted *FMR1* gene, it's the only one they have. Females always have that second *FMR1* gene, which may be able offset the effects of their impacted *FMR1* gene.

The Prevalence of Fragile X Syndrome

FXS has been detected in all populations and ethnic groups. As a result, efforts have been made to determine the overall prevalence of FXS and the difference in prevalence between males and females. The current agreed upon prevalence of FXS from the CDC is ~1 in 7,000 males and ~1 in 11,000 females.



While researchers do not have an exact number for how many Americans could have full-mutation Fragile X syndrome, the ratios noted above suggest that the raw number of individuals could be as high as 87,000 or as low as 38,000. Worldwide, the number could be between 1,400,000 and 777,000.

It is important to note that some published papers suggest greater prevalence and some lower prevalence than the numbers cited above.

Why is Fragile X less frequent and less severe in females?
The reason there are fewer females with FXS than males is that the gene for FXS is located on the X chromosome. Males, having only one X chromosome (XY), will develop FXS because there is a mutation of their single X chromosome. Females, who have two X chromosomes (XX), can have the unaffected X reduce the effects of the affected X. This typically leads to no or milder symptoms of FXS. (This is an important distinction, as many females with the full mutation do not consider themselves, nor are they considered by others, to have “Fragile X syndrome.”)

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Signs & Symptoms of Fragile X Syndrome

There are various signs and symptoms associated with FXS. No one individual will have all the features of FXS. The most common signs and symptoms include intellectual disability, behavioral and learning challenges, and various physical characteristics. These can be observed in both males and females with FXS, though there can be differences in how males and females present with FXS.

In Males

Behavioral characteristics can include ADHD, autism and autistic behaviors, social anxiety, hand-biting or flapping, poor eye contact, sensory disorders, sleep disorders, and increased risk for aggression.

Intellectual disabilities in FXS include a range from moderate learning disabilities to more severe intellectual disabilities. The majority of males with Fragile X syndrome demonstrate significant intellectual disability.

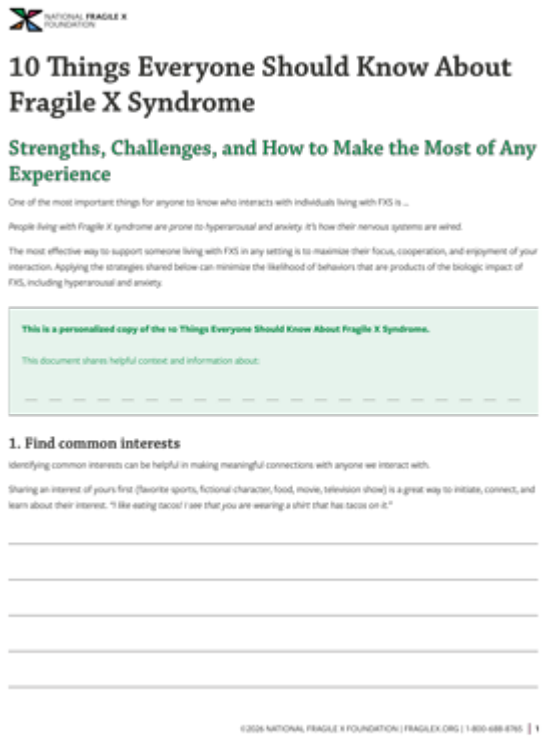
Other delays like speech, language, and motor delays (late crawling, walking, toileting) may also occur.

Physical features may include large ears, long face, soft skin, and large testicles (called “macroorchidism”) in post-pubertal males. Connective tissue problems may include ear infections, flat feet, high arched palate, double-jointed fingers, and hyper-flexible joints. Some features, such as a long face and macroorchidism, are more common after puberty.

Medical issues experienced by those with FXS may include strabismus (crossed eyes) and seizures.

Disposition in individuals with FXS includes being very social and friendly. They have excellent imitation skills, a strong visual and long term memory, they like to help others, and are nice, thoughtful people, with a wonderful sense of humor.

Related Resource



10 Things Everyone Should Know About Fragile X Syndrome

Strengths, Challenges, and How to Make the Most of Any Experience

This is a personalized copy of the 10 Things Everyone Should Know About Fragile X Syndrome.

This document shares helpful context and information about:

1. Find common interests

Common Strengths of Individuals with Fragile X Syndrome
3 pages,
Updated 06/30/2025
People with Fragile X Syndrome (FXS) are unique individuals who bring warmth, creativity, and heart to the world around them. While FXS can come with challenges, it also comes with strengths. Here are just a few.
Download the PDF

In Females

Behavioral characteristics seen in males can also be seen in females, though females often have a milder presentation of the syndrome's behavioral, physical, and connective tissue features.

Intellectual disabilities in females with FXS range from none to significant. Severity of learning disabilities in females:

- About one third have none to very mild learning disabilities.
- About one third have mild learning disabilities.
- About one third have moderate to significant intellectual disabilities.

A small percentage of females who have the full mutation of the *FMR1* gene that causes FXS will have no apparent signs of the condition — intellectual, behavioral, or physical. These females are often identified only after another family member has been diagnosed.

Additional challenges may include emotional and mental health issues, general anxiety, and social anxiety.

Learn more about females and FXS

Related Treatment Recommendations

Language in Fragile X Syndrome

What does language development look like for individuals with FXS? We discuss receptive language (what is understood), expressive language (how an individual communicates), pragmatics (how language is used), and speech (how sounds and words are produced).

Go to the treatment recommendation

Understanding Autism Spectrum Disorder in Fragile X Syndrome

Clarification on how the ASD diagnosis and FXS do and do not overlap. Understanding this distinction can be particularly helpful for genetic counseling and deciding on the most appropriate interventions.

Go to the treatment recommendation

Sensory Processing and Integration Issues in Fragile X Syndrome

Sensory-based hyperarousal is the most prevalent, troubling, and defining characteristic in Fragile X. Learning to manage hyperarousal proactively allows people to grow into themselves, not out of the problem.

Go to the treatment recommendation

Sleep in Children With Fragile X Syndrome

Sleep problems can be more frequent in children with developmental disabilities, including Fragile X syndrome. Ongoing support can play an important role. Depending on the presentation and primary disorder, treatment may include behavioral, pharmaceutical, and surgical interventions.

Go to the treatment recommendation

Physical Problems in Fragile X Syndrome

Multiple associated physical problems can occur with FXS, mostly related to loose connective tissue.

Go to the treatment recommendation
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Testing & Diagnosis

The *FMR1* DNA Test (sometimes called the Fragile X DNA Test) is considered the standard of care for determining the presence of Fragile X. DNA testing detects more than 99% of individuals (both males and females) with FXS and the Fragile X premutation.

Learn more about testing and diagnosis

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Genetics & Inheritance

Fragile X is passed down in families through a mutation in the *FMR1* gene located on the X chromosome. Everyone has an *FMR1* gene, the difference between having Fragile X and not having Fragile X comes down to CGG repeat number.

Fragile X-associated conditions include a wide range of physical, intellectual, and behavioral symptoms that can affect family members in many different ways. These

conditions are passed down in families through expansions of the *FMR1* gene.

FMR1 inheritance can be complex and confusing.

Learn more about genetics and inheritance

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Treatment & Intervention

While there is currently no cure for FXS, there are treatments and interventions that can improve the lives of individuals living with FXS and their families. Many families choose a combination of treatment interventions that may include medications and non-medication-based treatments, behavioral and other interventions, and school supports. What works for one person may not work for another. With the right combination, all individuals with FXS can make progress!

Medication Treatments

There are Fragile X clinics across the United States and around the world. Clinics can guide parents to medication options that may address the symptoms of FXS while new medications that aim to treat the underlying condition are coming on the market. Individuals with FXS may also be prescribed medications that treat specific symptoms of FXS, like ADHD or anxiety.

Learn more about medications and FXS

Related Treatment Recommendations

Medications for Individuals with Fragile X Syndrome

Medications are, at times, helpful to facilitate the individual’s ability to attain optimal life skills and allow for better integration into educational, adult, and social environments.

Go to the treatment recommendation

Behavioral Treatments

Individuals with FXS may display behaviors—some good and some bad—just like everyone else. It is possible that you may encounter challenging behaviors. Challenging behaviors can be addressed by assessing the problem behavior and creating a plan that includes appropriate support and accommodations. These steps will likely foster positive outcomes and set up the individual with FXS to succeed in their home, school, or community.

Learn more about managing behavior

Related Treatment Recommendations

Behavioral Challenges in Fragile X Syndrome

The most common behavioral challenges seen in FXS include those associated with generalized anxiety, social interaction difficulties, ADHD, self-injury, and aggression.

Go to the treatment recommendation

Educational Supports and Interventions

Most individuals with FXS qualify for special education services while in school. Education can be complemented by a variety of therapies that help individuals with FXS become more independent in the transitions from childhood through adolescence and into adulthood.

Learning does not end when school ends; the NFXF provides resources and tools to support individuals and their families through transitions from adolescence to adulthood.

Learn more about learning and school

Related Treatment Recommendations

Middle and High School Educational Recommendations for Children with Fragile X Syndrome

By using legal guidelines such as IDEA and implementing promising vocational,

educational, and life skills training practices, students with FXS can be better prepared for a successful transition into adulthood.

Go to the treatment recommendation

General Educational Recommendations for Students with Fragile X Syndrome

A basic framework for understanding different aspects of the educational system and an overview of the terminology. We also have resources for each level of the education system.

Go to the treatment recommendation

Elementary School Educational Recommendations for Children with Fragile X Syndrome

Information on legal educational policies and recommendations, plus strategies and supports that have proven successful for academic and adaptive functioning.

Go to the treatment recommendation

Early Childhood Developmental and Educational Guidelines for Children with Fragile X Syndrome

For all children within the early childhood age range of birth to 5 years and especially for young children with identified disabilities associated with a diagnosis like Fragile X syndrome (FXS), inclusive, nurturing, and developmentally appropriate environments and caregiving are essential to growth and development.

Go to the treatment recommendation

Learn More About Fragile X Syndrome

Browse topics or visit one of the following:

Adulthood
Autism
Caregiving

Daily Living Strategies
Females

Learning & School
Managing Behavior
Medication
Physical & Mental Health
Puberty & Sexuality



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