What is Fragile X Syndrome?

Fragile X syndrome (FXS) is a genetic disorder. FXS is caused by changes in a gene called Fragile X Messenger Ribonucleoprotein 1 (*FMR1*). *FMR1* usually makes a protein called FMRP that is needed for brain development. People who have FXS do not make this protein. Those with fragile X-associated disorders have changes in the *FMR1* gene, but usually still 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 a review of research studies estimated that about 1 in 7,000 males about 1 in 11,000 females have been diagnosed with FXS. [1]

Learn how FXS is inherited »

COVID-19: Information for parenting children with FXS

Learn more

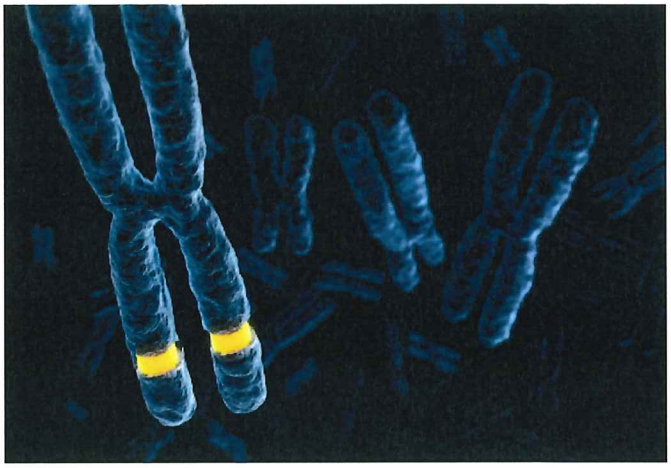
Signs and Symptoms

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 that can range from mild to severe. Females with FXS can have normal intelligence or some degree of intellectual disability. Autism spectrum disorder (ASD) also occur more frequently in people with FXS.

Testing/Diagnosis



FXS can be diagnosed by testing a person's DNA from a blood test. A doctor or genetic counselor 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.

Uncover the Facts: Fragile X Myth Busters for families and health professionals.

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, they 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 early intervention »

What to do if you think your child might have FXS

Local public school systems can provide services and support for children age 3 years and older. Children can access some services even if they do not attend public school. When parents are concerned about a child's development, it can be very challenging for them to figure out the right steps to take. States have created parent centers. These centers help families learn how and where to have their children evaluated and how to find services. For information about services in your state, you can access your state's parent center .

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 to get information about treatments, educational strategies, therapies and intervention.
- Connect with a Community Support Network (CSN) at the National Fragile X Foundation. CSNs are organized and run by parent volunteers and provide support to families.

CDC's Work on Fragile X Syndrome

CDC is working to learn more about the natural history of fragile X so that better approaches to intervention can be developed.

As part of this effort, CDC:

- Supported the National Fragile X Foundation to develop the Fragile X Online Registry With Accessible Research Database (FORWARD) . The purpose of FORWARD is to learn more about
  - Other conditions that commonly occur along with FXS,
  - The impact on the day-to-day lives of individuals living with FXS and their families,
  - Short-term and long-term outcomes, and
  - What type of interventions and support are most effective for different individuals and their families.
- Collaborated with the American Academy of Pediatrics to develop and distribute educational materials to healthcare professionals and families. These materials are designed to raise awareness of FXS and encourage early diagnosis so that people with FXS can receive appropriate care and services.
- Is working to learn more about how children with FXS develop compared to children with other conditions including ASD or developmental disability, and to learn more about children with FXS who also have other diagnosed conditions.

Other Resources

Fragile X Online Registry With Accessible Research Database (FORWARD)
Funded by CDC, FORWARD is the largest source of data on people with fragile X syndrome and their families.

FRAXA Research Foundation
FRAXA's mission is to accelerate progress toward effective treatments and ultimately a cure for fragile X by directly funding the most promising research. FRAXA also supports families affected by fragile X.

National Institutes of Health, Office of Rare Diseases Research, Genetic and Rare Diseases Information Center
The Office of Rare Diseases Research (ORDR) answers questions about rare diseases for patients, families, healthcare providers, researchers, students and educators. The ORDR website provides information about National Institutes of Health-sponsored biomedical research, scientific conferences, and rare and genetic diseases.

References

1. Hunter J, Rivero-Arias O, Angelov A, Kim E, Fotheringham I, Leal J. Epidemiology of fragile X syndrome: a systematic review and meta-analysis. Am J Med Genet A. 2014 Jul 164A(7): 1648-58.

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