

Facts About Fragile X Syndrome

What is fragile X syndrome?

Fragile X syndrome (FXS) is the most common known cause of intellectual disability that can be inherited, which means it can be passed from parent to child. FXS affects both males and females. However, females often have milder symptoms than males. The exact number of people who have FXS is not known, but a review of research studies estimated that about 1 in 7,000 males and about 1 in 11,000 females have been diagnosed with FXS.

What causes FXS?

The cause of FXS is genetic. FXS occurs when there is a change in the Fragile X Messenger Ribonucleoprotein 1 (*FMR1*) gene on the X chromosome. The *FMR1* gene makes a protein needed for normal brain development. In FXS, since 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. Some people with FXD may have noticeable symptoms, and others may not. 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 disorder (ASD) occurs 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 thinks 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 Center for Parent Information and Resources at www.parentcenterhub.org/find-your-center/.

In addition, CDC has links to information for families at www.cdc.gov/fragilex.

Additional resources include the National Fragile X Foundation (www.fragilex.org) and the FRAXA Research Foundation (www.FRAXA.org).

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. 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/fragilex