

# 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.