You may not have heard of fragile X–associated disorders, but they are the most common inherited cause of intellectual disabilities. Fragile X syndrome is caused by the full mutation in the *FMR1* gene.

People with fragile X syndrome don't have enough of a protein that is important to brain development. This is because they inherited an altered gene.

Fragile X syndrome in boys causes a range of intellectual disabilities, behavioral characteristics, and physical features. This guide will give you information about genetics, symptoms, and testing. Prenatal diagnosis can help you make decisions that are in the best interests of you and your family.

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For more in-depth information about fragile X syndrome, the following resources are available:

The National Fragile X Foundation
FragileX.org

The FRAXA Research Foundation
FRAXA.org

The March of Dimes
MarchOfDimes.org/Baby/Fragile-X-Syndrome.aspx

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Expectant parents:

What you need to know about fragile X disorders

A prenatal guide to genetics and testing

What does it mean to have fragile X syndrome?

Fragile X syndrome causes a range of intellectual disabilities and behavioral characteristics. While both boys and girls may have symptoms, boys are more likely to have fragile X syndrome.

Possible features associated with fragile X syndrome
(from the National Fragile X Foundation)

Physical

- Large ears
- Long, narrow face
- Prominent forehead or chin
- Low muscle tone
- Hyperflexible joints, particularly of the hands and wrists

Cognitive/behavioral

- Intellectual disability
- Hand-flapping and/or biting
- Poor eye contact
- Increased sensitivity to sounds, touch, crowds, and certain foods and textures

How is fragile X syndrome inherited?

Fragile X syndrome is caused by an altered gene.

Parents pass along genes to their children that determine everything about them from eye color to blood type. Parents cannot control what genes their children will inherit.

Genes are found on chromosomes. Girls have two "X" chromosomes and boys have one "X" and one "Y" chromosome. Fragile X syndrome is an X-linked disorder because the gene (FMR1) is found on the X chromosome.

Boys inherit fragile X syndrome from their mothers but girls can inherit it from either parent.

Fragile X syndrome is passed from mother to son because boys receive only a Y chromosome from the father. Girls receive one X chromosome from their mother and one from their father. However, paternal inheritance of fragile X syndrome is very rare.

The altered gene can be passed along for generations before a child actually has fragile X syndrome. Many women who carry the gene have no symptoms.

Boys tend to be more severely affected by fragile X syndrome than girls.

Because girls have two X chromosomes, they are most often less severely affected by fragile X syndrome. Chances are, one of the X chromosomes will not have a mutation and can make up for the altered gene.

Children with fragile X syndrome do live a normal life span although their challenges continue through adulthood.

Family history can indicate whether testing should be considered.

Since fragile X syndrome is inherited, there may be characteristics in your family that indicate whether your child might be at risk for the disorder. Do any relatives have:

- Intellectual disabilities, autism spectrum disorders, or other behavioral disorders?
- Female infertility or early or premature menopause?
- Late (after 50) onset neurological findings, including tremors, memory loss, or personality or psychiatric changes?

What testing is available for fragile X syndrome?

A simple blood test can help evaluate your risk of having a child with fragile X syndrome.

The XSense® blood test gives women information about their own genes. Depending on what the test reveals, your healthcare provider can estimate the chances that your child has the altered gene.

Additional prenatal diagnostic testing can provide a definitive answer.

Amniocentesis and chorionic villus sampling (CVS) can determine whether your developing baby inherited the altered gene. Since there is a risk of miscarriage associated with both tests, you should talk to your healthcare provider.

What happens after testing?

Knowing is the first step to understanding.

If testing indicates that your child may have fragile X syndrome, nothing can be done to change the baby's genetic makeup. However, knowing early will give you time to seek out information and support.

While there is no cure, early intervention and occupational and speech therapy can help children with fragile X syndrome lead full lives.

Ask your healthcare provider about genetic counseling.

If testing indicates that your baby may have or does have fragile X syndrome, genetic counseling will help you learn more about your risk and your baby's condition. Please speak to your healthcare provider.

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