

What is Fragile X syndrome?

Fragile X is a genetic syndrome that causes speech and learning delays, including autism. Fragile X syndrome happens more in males, but females can also be affected.

Why is my child being tested?

It is common to test children that have speech or learning delays for Fragile X syndrome. This includes children that have autism. Most children who are tested for Fragile X syndrome will have normal results- up to 5% of children tested will have a positive test result for Fragile X.

What causes Fragile X Syndrome?

We all have genetic information in every cell of our bodies. This genetic information is called DNA. DNA is held on structures called chromosomes and controls how our body develops and works. Fragile X is caused by extra repeating letters of DNA on the X chromosome. This extra genetic information, repeated letters of DNA, causes an important gene, called FMR1 to not work properly. When the FMR1 gene does not do its job, the body does not receive information that is needed in order for brain cells to work correctly. We all have a few of these extra letters, but when there are too many (over 200), we see the symptoms of Fragile X.

What does the test look for?

To test for Fragile X, we draw blood or saliva. Once the blood has been taken, the test looks for extra letters on the X chromosome. The test counts the number of repeats. If the number of repeats is long enough, FMR1 will be turned off and brain cells will be affected.

What are possible test results?

Positive test result: A positive test result means that there are over 200 extra repeats on the X and the child has Fragile X. A family with Fragile X syndrome should schedule a meeting with genetic services.

Premutation: This means that there are anywhere from 50 to 200 extra repeats on the X chromosome. This child does not have Fragile X syndrome, but could be at risk for other health issues: ovary issues in women and tremors in men. Women with premutations can have children with Fragile X syndrome, because repeats may get larger when passed on from mother to child. A family with this diagnosis should schedule a meeting with genetics services.

Intermediate: This means that there are 45-54 repeats, which are a few extra but not very much. There are no health or development issues in a carrier of intermediate repeats. But, this means that a female with intermediate repeats can have children who are premutation carriers.

Negative Test result: This means that there are 50 repeats or less on the X chromosome. With 50 repeats or less, the gene on the X does not get turned off. The gene stays on and working. The child does not have Fragile X syndrome. If there are questions, you may meet with genetics.



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Fragile X Syndrome/ FMR1 Related Disorders

Negative

- <50 extra repeats on the X chromosome
- Child does **NOT** have Fragile X syndrome
- No expected health or reproductive issues
- If you have questions, schedule appointment with Genetics

Intermediate

- 45-54 extra repeats on the X chromosome
- Child does **NOT** have Fragile X syndrome
- No expected health issues
- Females may have children that are Premutation carriers
- Schedule appointment with Genetics

Premutation

- 55 to 200 extra repeats on the X chromosome
- Child does **NOT** have Fragile X syndrome
- Females may have children with Fragile X syndrome
- Increased chance of some health concerns
- Schedule appointment with Genetics

Positive

- > 200 extra repeats on the X chromosome
- Diagnostic for Fragile X syndrome
- Symptoms vary from person to person
- Girls may pass this on to their children
- Schedule appointment with Genetics