



WHAT IS FRAGILE X?

Fragile X is a group of disorders that can affect individuals across generations.

FRAGILE X INCLUDES:



Fragile X Syndrome (FXS)

A genetic condition that causes intellectual disability, behavioral and learning challenges and various physical characteristics. Though FXS occurs in both genders, males are more frequently affected than females, and generally with greater severity. It is also the most common known single gene cause of autism spectrum disorder.



Fragile X-associated tremor/ataxia syndrome (FXTAS)

A neurological disorder that causes tremors and changes to balance, memory, mood, and intellect in some adults over the age of 50.



Fragile X-associated primary ovarian insufficiency (FXPOI)

A disorder that causes irregular menstrual cycles, infertility, and premature menopause in some females of reproductive age.

Facts About Fragile X

- The gene responsible for Fragile X is called FMR1, which is found on the X chromosome. Changes in the FMR1 gene are what lead to Fragile X.
- Fragile X can be accurately diagnosed with a simple blood test called the FMR1 DNA test.
- The test must be ordered by a genetic counselor or physician.
- An estimated 1.5 million Americans are carriers of a Fragile X mutation.
- Approximately 100,000 individuals in the U.S. have FXS, many of them undiagnosed.
- Both sexes can have FXS. Females with FXS often have milder symptoms than males.
- People with FXS can have anxiety, behavioral issues, and language delays that manifest in early childhood.
- Other characteristics of FXS can include poor eye contact, hand-flapping, flexible joints, long face, and prominent ears.
- Fragile X premutation carriers have what is called a premutation of the FMR1 gene; they do not have Fragile X syndrome.
- Some, but not all, premutation carriers will develop FXTAS or FXPOI.
- FXTAS occurs more commonly in males who are premutation carriers, though some females also develop some features of FXTAS.
- Other issues sometimes seen in premutation carriers include high blood pressure, migraine headaches, depression, anxiety, hypothyroidism, fibromyalgia, and sleep apnea.





HOW IS FRAGILE X INHERITED?

- Both males and females can be Fragile X premutation carriers.
- Male premutation carriers pass their Fragile X gene to all their daughters, who will be premutation carriers, but to none of their sons.
- Female premutation carriers have a 50 percent chance with each pregnancy of passing the Fragile X mutation on to a son or daughter, some of whom will have FXS.



WHAT ARE THE TREATMENTS FOR FRAGILE X?

Treatment for FXS consists primarily of special education and speech and language, occupational, and physical therapies. Medications can also be helpful.

Many therapeutic activities and medications can also help those with FXTAS and FXPOI. Currently, there is no cure for FXS, FXTAS, or FXPOI, though many symptoms can be reduced.



WHAT IS THE NATIONAL FRAGILE X FOUNDATION?

The National Fragile X Foundation (NFXF) is a 501(c)(3) nonprofit charitable organization dedicated to raising awareness, supporting research, and aiding families affected by Fragile X.

The NFXF provides:

- Toll-free telephone and email support.
- Referrals to the nearest Community Support Network group, which is typically a parent-led support group for families and caregivers.
- Referrals to a network of nationally recognized Fragile X clinics.
- Legislative advocacy and financial support for research including the translation of science into new and improved treatments.

If you or someone you know lives with Fragile X, please contact the NFXF. We can help!



**NATIONAL FRAGILE X
FOUNDATION**

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