FACT SHEET | NATIONAL **FRAGILE X** FOUNDATION
— TENNESSEE —

What is Fragile X?

Fragile X is a group of conditions associated with alterations in the *FMR1* gene on the X chromosome.

Fragile X Syndrome (FXS)

FXS is an inherited genetic condition caused by a full mutation of the *FMR1* gene that may cause intellectual disability, behavioral and learning challenges, and various physical characteristics. It is the leading, known inherited cause of intellectual disability and autism spectrum disorder.

Fragile X-associated Primary Ovarian Insufficiency (FXPOI)

FXPOI is a condition in which the ovaries are not functioning at full capacity in a woman with the *FMR1* premutation that can cause infertility, early menopause, and other ovarian issues. FXPOI occurs in about 20% of women with the *FMR1* premutation.
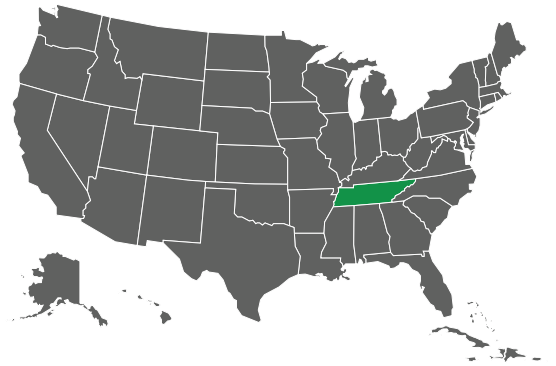
Fragile X-associated Tremor/Ataxia Syndrome (FXTAS)

FXTAS is an adult-onset neurodegenerative disorder, more common in males than females with the *FMR1* premutation. FXTAS is associated with tremors, balance problems, and other neurological signs. The risk of developing FXTAS in any given individual is influenced by their CGG repeat size, sex, and age.

What conditions benefit from Fragile X research?

- Alzheimer's Disease
- Parkinson's Disease
- Down Syndrome
- Autism
- Cystic Fibrosis
- Polycystic Ovarian Syndrome
- Epilepsy
- Amyotrophic Lateral Sclerosis

How common is Fragile X?

	United States	Tennessee
Fragile X Premutation	~1.5 million	~31,925
Fragile X Syndrome	~100,000	~2,150

Is Fragile X rare?

FXS is considered a rare disease. Rare diseases are defined as less than 200,000 individuals in the U.S.

The Fragile X premutation is not rare.

How is Fragile X diagnosed?

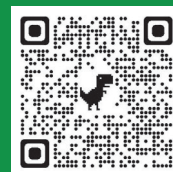Through a simple *FMR1* / Fragile X DNA test.

Is there a cure?

Not yet.

Researchers are working on gene therapies as long-term treatment options. There are also several interventions in clinical trials.

Until they are successful, doctors can only treat the symptoms of the Fragile X conditions and disorders with approved interventions.

Want to know more?

- fragilex.org
- contact@fragilex.org