



## 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.



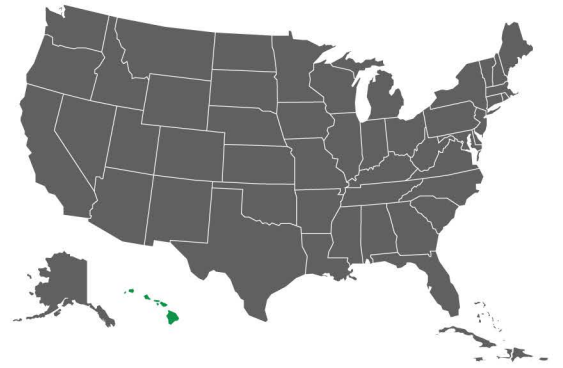
## Fragile X-associated Tremor/Ataxia Syndrome (FXTAS)

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 **Hawaii**

<b>Fragile X Premutation</b>	~1.5 million	~6,450
<b>Fragile X Syndrome</b>	~100,000	~450



## 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](http://fragilex.org)
- [contact@fragilex.org](mailto:contact@fragilex.org)