

FACT SHEET

Puerto Rico

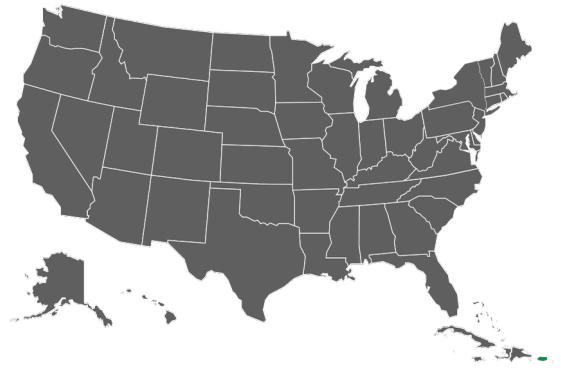


NATIONAL FRAGILE X
FOUNDATION



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 cause of inherited intellectual disability.



How common is Fragile X?

United States

Puerto Rico

Fragile X premutation \approx 1.5 million \approx 14,375

Fragile X syndrome \approx 100,000 \approx 975



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.



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.



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.



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.



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

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