What is Fragile X?

Fragile X is a group of conditions associated with alterations in the \textit{FMR1} gene on the X chromosome.

**Fragile X Syndrome (FXS)**

FXS is an inherited genetic condition caused by a full mutation of the \textit{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 \textit{FMR1} premutation that can cause infertility, early menopause, and other ovarian issues. FXPOI occurs in about 20% of women with the \textit{FMR1} premutation.

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

FXTAS is an adult-onset neurodegenerative disorder, more common in males than females with the \textit{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 common is Fragile X?**

- \textbf{United States} \\
  - \textbf{Fragile X Premutation} \( \sim 1.5 \text{ million} \) \\
  - \textbf{Fragile X Syndrome} \( \sim 100,000 \)

- \textbf{West Virginia} \\
  - \textbf{Fragile X Premutation} \( \sim 7,950 \) \\
  - \textbf{Fragile X Syndrome} \( \sim 550 \)

**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 \textit{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