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?**

<table>
<thead>
<tr>
<th>United States</th>
<th>Rhode Island</th>
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<tbody>
<tr>
<td>Fragile X Premutation</td>
<td>~1.5 million</td>
</tr>
<tr>
<td>Fragile X Syndrome</td>
<td>~100,000</td>
</tr>
</tbody>
</table>

**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