

# FACT SHEET

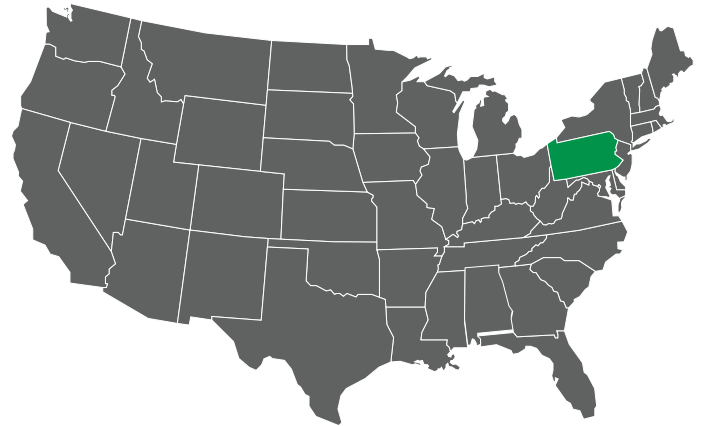


NATIONAL FRAGILE X FOUNDATION  
SOUTHEAST PENNSYLVANIA CHAPTER



## What is Fragile X?

Fragile X is a group of conditions associated with changes in the FMR1 gene on the X chromosome. The changes can cause a “premutation” or a “full mutation,” which can result in a Fragile X-associated disorder.



## How common is Fragile X?

### U.S.

~1.5 million carriers  
~100,000 with FXS

### PENNSYLVANIA

~59,000 carriers  
~4,000 with FXS



## Fragile X Syndrome

FXS is a genetic condition caused by a full mutation of the FMR1 gene. Symptoms include intellectual disability, behavioral and learning challenges, and various physical characteristics. It is also the most common known single gene cause of Autism Spectrum Disorder.



## Fragile X-associated Primary Ovarian Insufficiency

FXPOI occurs in ~22% of female carriers and is a cause of infertility, early menopause, and other ovarian problems.



## Fragile X-associated Ataxia/Tremor Syndrome

FXTAS is an adult onset (age 50+) condition that causes neurological and psychiatric symptoms in carriers. It is commonly misdiagnosed as Parkinson's.



## What conditions benefit from Fragile X research?

- Alzheimer's Disease
- Parkinson's Disease
- Down Syndrome
- Autism Spectrum Disorders
- Cystic Fibrosis
- Polycystic Ovarian Syndrome
- Epilepsy
- ALS



## How is Fragile X diagnosed?

Through a simple DNA blood test.



## Is there a cure?

Not yet. Nor is there a treatment specifically indicated for Fragile X.

Researchers are working on gene therapies, protein synthesis for longterm treatments, and other possible drug and non-drug treatments. Doctors currently use off-label medications and other interventions.



## Want to know more?

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