## FACT SHEET OKLAHOMA OKLAHOMA 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 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 Oklahoma

Fragile X Premutation ~1.5 million ~18,175 Fragile X Syndrome ~100,000 ~1,225



#### 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