

National Fragile X Center at Emory

*promoting excellence in
clinical care,
research and
education*



We have an exciting new study to discover genes that affect the risk and severity of three fragile X-associated disorders.

Our goal is to find new avenues for potential treatments.

National Fragile X Center at Emory

Center Director:

Stephen T. Warren, PhD

Center Co-Director:

Stephanie Sherman, PhD

Additional sites:

University of Colorado Denver

Maureen Leehey, MD

Please contact us

if you have any questions or would like additional information.

Krista Charen

404-778-8479

krista.charen@emory.edu

Lisa Shubeck

404-778-8478

Lshubec@emory.edu



National Fragile X Center

Excellence in Clinical Care, Research, and Education

Modifiers of Fragile X-Associated Disorders (FX-MOD)

...tracking down
genes that interact with
FMR1



Help advance fragile X research by participating in any of our three new studies!

Study 1: Modifiers of FXPOI in women with the premutation

Fragile X-associated primary ovarian insufficiency (FXPOI) leads to reduced ovarian function and sub-fertility. It affects about 20% of women with the premutation. Our goal is to discover genes that modify the risk and severity of FXPOI.

Study Groups:

- Group 1: females with premutation, age 18-75, with early symptoms of FXPOI
- Group 2: females with premutation, age 50-75, with age at menopause at 50 years or older

Study Activities:

- Medical history review
- Blood or saliva sample
- Health and well-being questionnaires

Participants will receive a \$25.00 gift card for a completed blood or saliva sample.

Study 2: Modifiers of FXTAS in individuals with the premutation

Fragile X-associated tremor ataxia syndrome (FXTAS) is a neurological disorder that affects about 40% of older men and about 15% of older women who carry the premutation. Our goal is to discover genes that modify the risk and severity of FXTAS.

Study Groups:

- Group 1: individuals with premutation, age 50-90, with early symptoms of FXTAS
- Group 2: males with premutation, with no symptoms of FXTAS before the age of 70

Study Activities:

- Medical history review
- Some will visit Emory for a neurological exam
- Blood or saliva sample

Participants will receive a \$25.00 gift card for a completed blood or saliva sample.

Study 3: Modifiers of seizures in individuals with FXS

Seizure disorders affect about 15% of children with fragile X syndrome (FXS) and can lead to increased severity of symptoms. Our goal is to discover genes that modify the risk for seizures in individuals with FXS.

Study Groups:

- Group 1: individuals with FXS, age 4-50, with at least 1 seizure
- Group 2: males with FXS, age 17-50, with no history of seizures

Study Activities:

- Medical history review
- Blood or saliva sample

Participants will receive a \$25.00 gift card for a completed blood or saliva sample.