



EMORY  
UNIVERSITY  
SCHOOL OF  
MEDICINE

## 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



## National Fragile X Center at Emory

Promoting excellence in clinical  
care, research and education

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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.



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## National Fragile X Center at Emory

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### PLEASE CONTACT US

If you have any questions or would like  
additional information.

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Child Health and Human Development. It has  
been approved by the Emory University Institu-  
tional Review Board, Study ID 00074941.

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

### **Study 1: Modifiers of FXPOI in women with the premutation**

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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**

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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**

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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.