

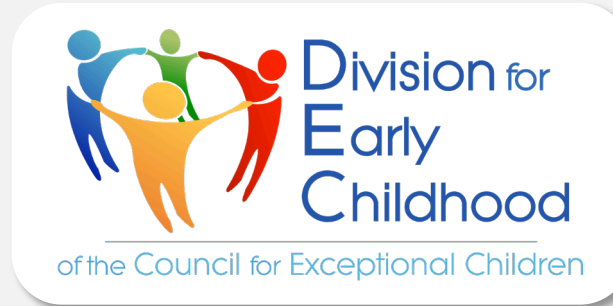


# **FRAGILE X and EARLY INTERVENTION:**

## **Increasing the Diagnosis and the Participation of Historically Underserved Populations**



# Aligned Professional Competencies and Frameworks



## Division for Early Childhood (DEC) RPs

**F2:** Practitioners provide the family with up-to-date, comprehensive and unbiased information in a way that the family can understand and use to make informed choices and decisions.

# Aligned Professional Competencies and Frameworks



## Knowledge & Skills for Service Coordinators (KSSC)

### **1.1: Infant/Toddler Development**

Explain developmental delays and disabilities and their impact on children's development and learning to support intervention decisions.

# Aligned Professional Competencies and Frameworks



## Pyramid Model Framework (PMF) Key Practices



**Building Partnerships  
with Families**

# Presenters



**Robert Miller**



**Amie Milunovich**



# NATIONAL FRAGILE X FOUNDATION

Our mission is to provide unwavering support for every family affected by Fragile X, while relentlessly pursuing a cure.



[fragilex.org](http://fragilex.org)

# What is Fragile X?

Fragile X is an inherited genetic disorder affecting the X chromosome in both males and females. The mutation can lead to:

**Fragile X  
syndrome**

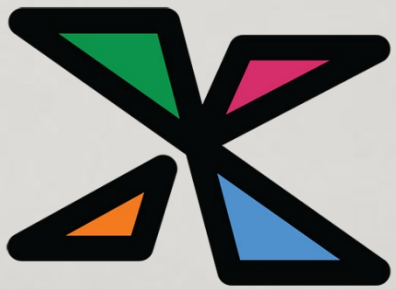
**Fragile X-  
associated  
tremor-ataxia  
syndrome  
(FXTAS)**

*Adult-onset degenerative  
neurological condition*

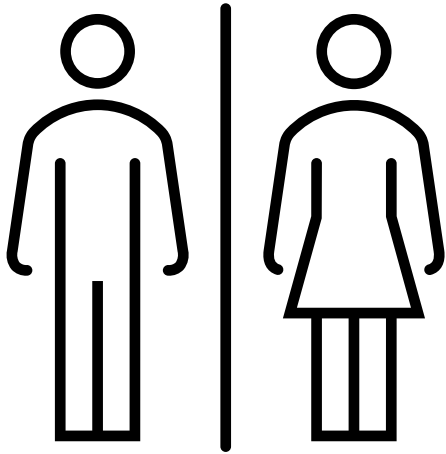
**Fragile X-  
associated  
primary ovarian  
insufficiency  
(FXPOI)**

*Reproductive issues including  
early menopause*

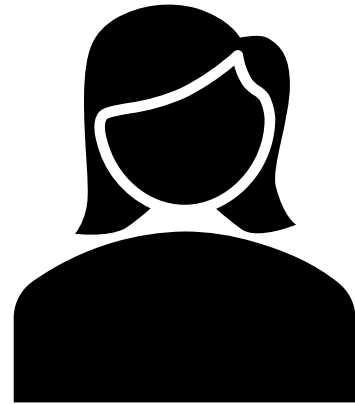
**Other  
premutation  
carrier issues**



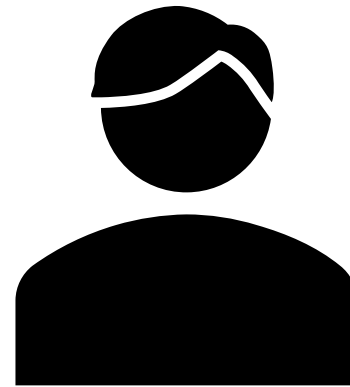
# HOW COMMON IS FRAGILE X?



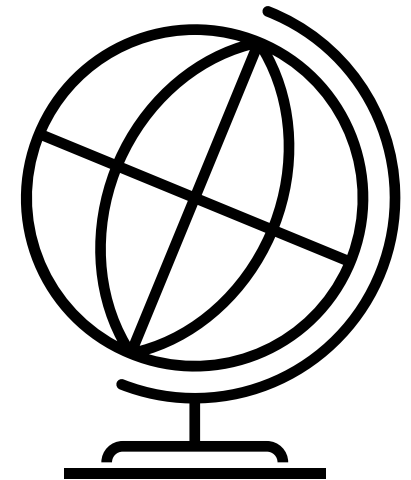
FXS affects 1 in 3,600 males and 1 in 4,000- 6,000 females



1 in 151 women are carriers in the US



1 in 468 men are carriers in the US



Fragile X appears in all world populations





Approximately 50–100,000 Americans have Fragile X syndrome, and many hundreds of thousands are carriers who, in addition to potentially passing the mutation to their offspring, are, themselves, at risk for having or developing a Fragile X condition.



# What is the NFXF asking of Early Intervention Providers?



**Increased awareness about Fragile X by Early Intervention providers.** *The NFXF can provide informational materials!*

**Information sharing about Fragile X treatment clinics with families who may have a child already diagnosed with FXS.** *Clinics exist throughout the US!*

**Information sharing with families who have a child with developmental delay or intellectual disability for which the cause has not been determined.** *The Fragile X DNA test is better than 99% accurate!*

**Information sharing with families who have a child diagnosed with autism or who is said to have an autism spectrum disorder.** *For some children who are diagnosed with autism or ASD (a behavioral diagnosis) the medical cause is FXS!*

- Approximately  $\frac{1}{3}$  to  $\frac{1}{2}$  of all children with fragile X syndrome have some degree of autistic behavior.
- For between 2% and 6% of children with *classic* autism, the cause of their autism is fragile X syndrome.
- Autism is a behavioral diagnosis while fragile X syndrome is a medical diagnosis. It is not uncommon for a child to initially be diagnosed with ASD and later to receive an additional diagnosis of FXS or vice versa.

A close-up photograph of a baby's face, smiling broadly, showing teeth. The baby has dark hair and is wearing a dark-colored garment with a patterned collar. The background is a soft-focus outdoor scene with a body of water and a distant shoreline under a hazy sky.

# Autism and Fragile X

## Cognitive Issues



## Sensory Processing Issues



Developmental Delay/Intellectual Disability including:

- Short attention span
- Difficulty with auditory processing

Young children

- Excessive mouthing and drooling
- Difficult to calm and comfort

As they grow

- Mouth stuffing
- “Picky” eaters
- Sensitive to sounds – especially unexpected ones





# Behavioral Issues

- Hyperarousal
- Anxiety
- Hand flapping or biting
- Difficulty making eye contact
- Perseveration
- Hyperactive/short attention span
- Easily distracted
- Shyness
- Impulsiveness
- Hypervigilance
- Difficulty with transitions
- Tantrums/aggressiveness/self-injury

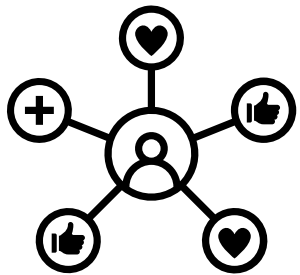


# Behavior Strengths



- Very social and friendly
- Excellent imitation skills
- Strong visual memory/long term memory
- Likes to help others
- Wants to please/make others happy
- Are nice, thoughtful people
- Wonderful sense of humor

*Girls, in general, are less affected than boys*





# Speech & Language

- Slower to learn to speak
- Perseveration
- Cluttered quality
- Tangential speech
- Echolalia
- Dysfluency/stuttering
- Rapid

## Strengths

- Quality improves with age
- Excellent mimic ability
- Receptive skills much better than expressive skills

# Gross & Fine Motor Skills

## Gross Motor

- Low muscle tone
- Challenges with hand/eye coordination



## Fine Motor/Self-Help

- Low muscle tone
- Hyper extensible finger joints
- Difficulties with self feeding, dressing, toileting



# Medical Issues

Ear Infections

Hernias

Cleft palate

Reflux

Heart  
conditions

Joint dislocation

Flat feet and  
pronated ankles

Vision problems

Scoliosis

Seizures

## **FMR1 DNA test (Fragile X DNA test)**

- **Standard of care for determining the presence of Fragile X.**
- **DNA testing detects more than 99% of individuals (both males and females) with FXS (the “full-mutation”), as well as Fragile X carriers (the “premutation”).**
- **The test requires a blood draw.**

*In families with a known Fragile X history, testing can be done preconception (i.e. of an adult who want to know if he or she is a carrier), prenatally or in infancy. However, it is more typically done in the preschool years or later as delays in development become more pronounced.*

## **How and When is Fragile X Diagnosed?**





# What is the Cost of Testing?

The cost for the DNA test for Fragile X can vary widely depending on insurance coverage, deductibles, the state of residence, etc.

Because of the many variables, the DNA test for Fragile X can range from under \$100 up to \$1000.

Families should check with their insurance company for costs and any requirements that need to be met.

**\*Note that the test may be covered by insurance, including Medicaid, and may be free or subject to copays/deductibles.**



# Why Diagnosis is Important

- Can lead to multiple immediate and extended family members learning they may have or *are at-risk for developing* a Fragile X disorder.
- Will lead to specific Fragile X evidence and [consensus-based treatment](#) including therapies, special education, counseling and medication.
- Allows for families to make the reproductive decisions best for their family. (Many families have two, three or more children with FXS.)
- Allows for families to be a part of the global Fragile X community of families and professionals. (The NFXF has parent-led groups in almost all 50 states.)
- Allows for those who are interested to become part of a Fragile X research project that can benefit their child, themselves and millions throughout the world!

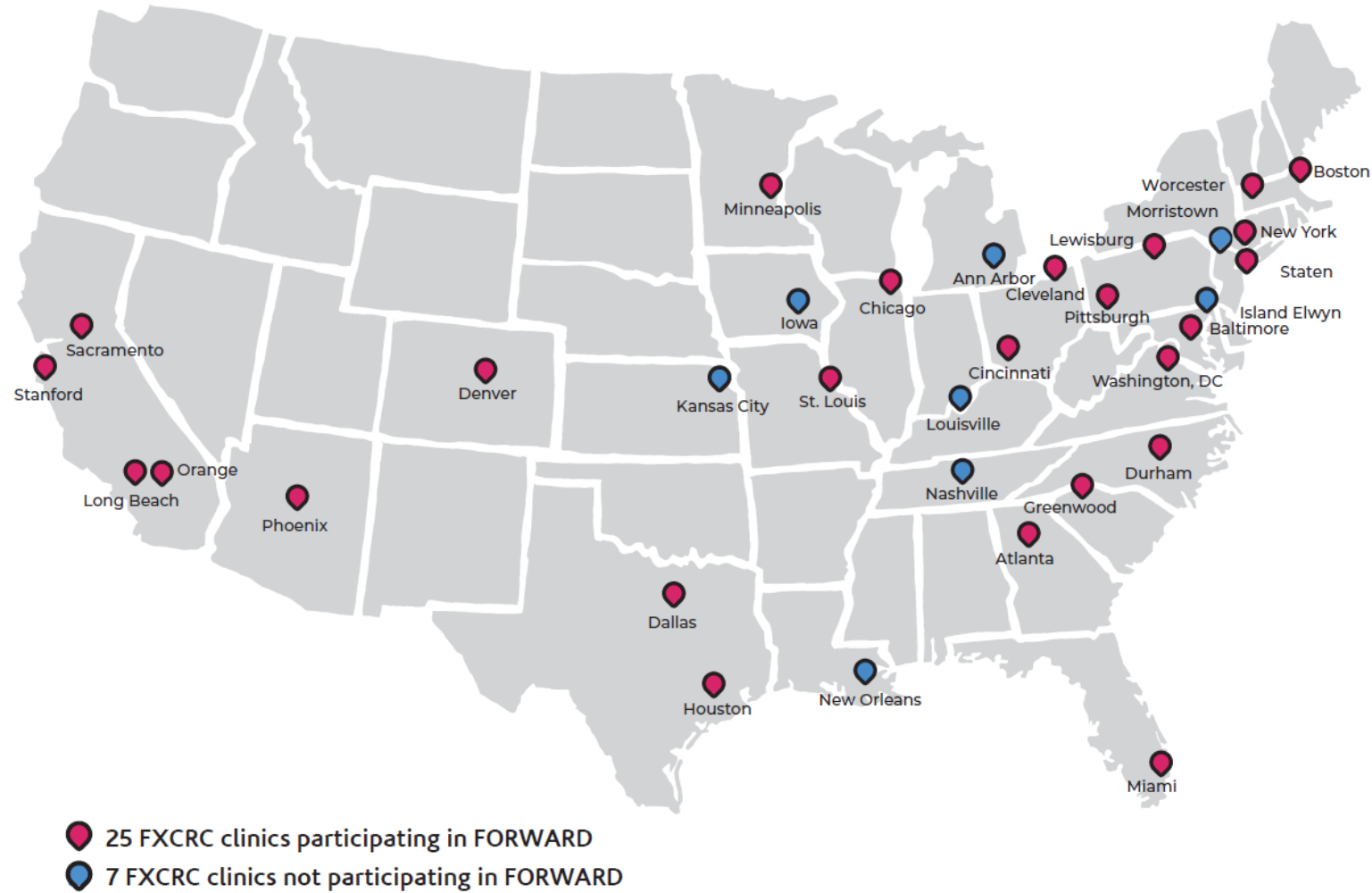
# What is the Fragile X Clinical & Research Consortium?

The Fragile X Clinical & Research Consortium (FXCRC) is a group of over 30 Fragile X specialty clinics at hospitals throughout the US and overseen by the NFXF. (It also includes international clinics.)

Fragile X clinics provide medical services, including medication evaluation and consultation, supervised by a physician and supported by the latest medical, educational, and research knowledge available. Services, such as genetic counseling and occupational, speech, language, and behavioral therapies are also available either at the clinic or by referral.

Many of the clinics participate in collaborative research efforts.

# Where are clinics located?



<https://fragilex.org/our-research/fragile-x-clinics/>

# Clinics Nearest to the State of Virginia

## Children's National Health System

### Children's National Fragile X Clinic

6833 4th Street NW

Washington, DC 20012

**PHONE:** [1-202-476-6249](tel:1-202-476-6249)

**EMAIL:** [cbouska@childrensnational.org](mailto:cbouska@childrensnational.org)

Sinan Omer Turnacioglu, MD, Medical Director

Cecilia Bouska, Clinic Coordinator

## Duke University Medical Center

### Lenox Baker Children's Hospital / Child Development Program

3000 Erwin Road

Durham, North Carolina 27705

**PHONE:** [1-919-668-0477](tel:1-919-668-0477)

**EMAIL:** [alicia.russell@duke.edu](mailto:alicia.russell@duke.edu)

Alice Bryn Salter, MD, Medical Director

Alicia Russell, Clinic Coordinator

## Cincinnati Children's Hospital Medical Center

### Cincinnati Fragile X Research & Treatment Ctr.

3333 Burnet Avenue

Cincinnati, Ohio 45229

**PHONE:** [1-513-803-1150](tel:1-513-803-1150)

**EMAIL:** [fragilex@cchmc.org](mailto:fragilex@cchmc.org)

Craig Erickson, MD, Medical Director

Austin Corsmeier, Clinic Coordinator

## Norton Children's Developmental Center & Genetics Ctr.

### The Norton Center Fragile X Clinic

411 East Chestnut Street, Level 2

Louisville, Kentucky 40202-3828

**PHONE:** [1-502-588-0850](tel:1-502-588-0850)

**EMAIL:** [jeanie.hammond@nortonhealthcare.org](mailto:jeanie.hammond@nortonhealthcare.org)

Joseph Hersh, MD, Medical Director

Jeanie Hammond, LCSW, Clinic Coordinator





Families are eligible to receive a grant of up to \$300 per family, per year for travel including accommodations, food, and/or childcare expenses. These funds can also be used to help pay for a clinic visit.

*These funds cannot be used for “testing for Fragile X.” The person must already have a diagnosis of fragile X syndrome or a premutation carrier.*

## Community Support Networks



- State-based parent contacts and volunteer-lead chapters and affiliates across the nation that provide local support, educational and awareness opportunities to families.



FRAGILE X ONLINE REGISTRY WITH  
ACCESSIBLE RESEARCH DATABASE

## Advancing FXS Research

FORWARD helps advance FX research by identifying individuals who may be eligible for a research study and connecting them to researchers.

## LONGITUDINAL DATABASE



Clinical and parent-reported data on individuals with full mutation Fragile X syndrome

[FORWARDFX.ORG](https://FORWARDFX.ORG)



• White non-Hispanic:	74.6%
• Black non-Hispanic:	8.1%
• Asian:	3.4%
• Hispanic:	12.6%
• Other	1.4%

# Race/Ethnicity Distribution of FORWARD Participants

(Total participants=1843)

# Why are Certain Populations underrepresented in FORWARD?

*Fragile X researchers and clinicians are not certain why some groups do not participate in Fragile X research such as FORWARD. The reason may be a combination of one or more of the following:*

- Lack of awareness about Fragile X therefore resulting in lack of diagnosis.

- Concern about the cost of getting tested and/or visiting a clinic.

- Lack of awareness about the existence of Fragile X clinics therefore resulting in not being informed of opportunity to participate in research.

- Skepticism about the benefits of research including skepticism based on historical abuses of minorities participating in medical research and misuse of research data.





- [FORWARD](#) is helping to reach an enhanced understanding of FXS.
- Provides evidence-based descriptions of how FXS presents itself across a person's lifetime including co-occurring conditions, associated risk factors, and service barriers and needs.
- [FORWARD has produced 26 Peer reviewed journal publications](#)





# NATIONAL FRAGILE X FOUNDATION

## WHERE TO FIND US



(202) 747-6203 /  
(800) 688-8765



[contact@fragilex.org](mailto:contact@fragilex.org)



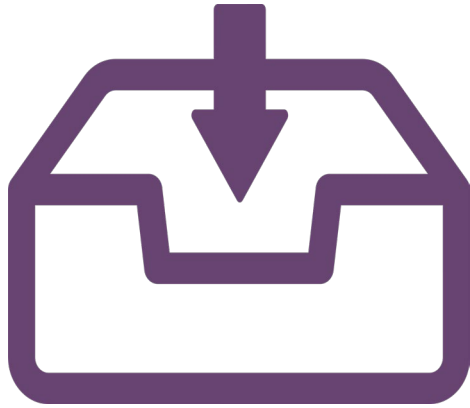
1012 14th Street NW,  
Suite 500,  
Washington, DC, 20005

FRAGILEX.ORG



Next session on  
**November 12th @ 12noon**

1



Check Your Inbox

2



Take the  
Survey

3



Download the  
Certificate!