

About Fragile X-associated Disorders

Fragile X is a family of conditions associated with changes in the Fragile X gene.

The gene (also known by its scientific name of “FMR1”) can be normal, but it can also exhibit a “premutation” or a “full mutation.” When one of these is present, it can result in a Fragile X-associated Disorder (FXD). These include:

- **Fragile X syndrome (FXS):** The most common known cause of inherited mental impairment, FXS affects intellectual, behavioral and social development. It occurs in both males and females who have a full mutation of the FMR1 gene, though its symptoms are generally more severe in males.
- **Fragile X-associated primary ovarian insufficiency (FXPOI):** A condition affecting ovarian function that occurs in some adult females who have a premutation of the FMR1 gene. (Females and males who have a premutation are also referred to as “carriers.”)
- **Fragile X-associated tremor/ataxia syndrome (FXTAS):** An adult onset neurological condition causing tremors, memory and balance problems. It occurs in some adult carriers (more commonly in males).

For more information prior to your clinic visit, log on to The National Fragile X Foundation’s website (www.FragileX.org), or call the foundation toll-free at 1-800-688-8765.

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About the NFXF

The National Fragile X Foundation was founded in 1984 to support individuals with fragile X syndrome, their families, and the professionals who work with them. Today, it is a comprehensive resource for all Fragile X-associated Disorders. The organization offers a toll-free phone line and email support; education through an extensive website, quarterly journal and other publications; and awareness activities throughout the country.

It also organizes international conferences, funds scientific research, and leads legislative advocacy efforts in Washington, D.C.



Contacts

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In addition, the NFXF organizes “LINKS Groups” that provide parent support and many other resources for families affected by Fragile X. Consult the listing below for more information.

Fragile X Association of New York (Brooklyn)

Anita Inz ■ Home: (718) 875-4901
E-Mail: anita.inz@gmail.com
Web: http://www.fragilex.org/html/new_york_city.htm

Fragile X Association of New Jersey

Paula Fasciano ■ Home: (732) 591-2497
E-Mail: thefascianos@gmail.com
Web: www.fragilex.org/html/new_jersey.htm

George A. Jervis Clinic Fragile X Center at the Institute for Basic Research in Developmental Disabilities



NYS Office For People With Developmental Disabilities

Putting People First



**Clinical & Research
CONSORTIUM**



The National **Fragile X** Foundation

Dedicated to Serving the Fragile X Community



The Institute for Basic Research Fragile X Center was founded in 2000. It is part of the Fragile X Clinical & Research Consortium (FXCRC), a group of independent clinics throughout the U.S. and into Canada and Mexico that are dedicated to serving the needs of families whose members are affected by one or more of the three identified Fragile X-associated Disorders (FXD): fragile X syndrome (FXS), fragile X-associated tremor/ataxia syndrome (FXTAS), and fragile X-associated primary ovarian insufficiency (FXPOI).

The FXCRC was founded by The National Fragile X Foundation (NFXF), which supports the formation of each clinic and helps them coordinate with others in a truly collaborative network that makes clinic services as accessible and convenient as possible.



The **Fragile X Center** is a component of the George A. Jervis Clinic at the Institute for Basic Research in Developmental Disabilities (IBR), the research arm of the New York State Office for People With Developmental Disabilities. The Jervis Clinic is a specialized diagnostic and research center, providing neurological, psychiatric, psychological, and genetic services to individuals with developmental disabilities, including fragile X syndrome (FXS).

FXS is a priority area of research at IBR. Institute scientists and clinicians have:

- Been participating in a multi-site project to facilitate the development and testing of new targeted drug treatments for FXS and autism
- Demonstrated the association of FXS with autism
- Investigated the relationships of the molecular findings of the fragile X mutation to the clinical and psycholinguistic manifestations of the disease and the degree of disability it causes
- Undertaken studies to identify the function of the Fragile X protein (which is absent in individuals with FXS) in model organisms, including knockout (KO) mice
- Pioneered in the development of laboratory diagnostics for FXS
- Had the most extensive experience in FXS prenatal diagnosis of any laboratory in the world

Medical Director: W. Ted Brown, MD, PhD

Clinic Coordinator: Nancy J. Zellers, MS, CGC



Ted Brown, MD, PhD
Medical Director

Range of Services

The **Fragile X Center** offers a broad spectrum of services to individuals who have or are suspected of having FXS, and to their families:

- Genetic evaluations and counseling
- Genetic and prenatal testing
- Medical evaluations, including:
 - Psychiatry
 - Medication review and recommendations
 - Neurology—including evaluations for adults with fragile X-associated tremor/ataxia syndrome (FXTAS)
- Psychological evaluations, including:
 - Speech and language
 - Neuropsychology
 - Behavioral support
 - Education (in school and home) and IEP review
- Other services, including:
 - Assistive technology
 - Educational programming and resource center
 - Outreach

Our staff is available to provide in-service training for teachers, therapists, and others working in the field of developmental disabilities who would like to learn more about the unique characteristics of FXS.

