

Fragile X Syndrome

TIPS AND RESOURCES FOR FAMILIES



What is Fragile X syndrome?

Fragile X syndrome is the most common inherited cause of intellectual disability. It is caused by a mutation of a gene (FMR1) on the X chromosome responsible for making a protein that plays a role in brain development. When the gene is fully mutated, it becomes inactive and does not make the protein. This lack of protein results in fragile X syndrome.

Some people may be premutation carriers and show no outward signs of fragile X, yet they may pass on the full gene mutation to their children. Approximately 1 in 3600 males and 1 in 4000 to 6000 females have fragile X syndrome.

What are the effects of Fragile X syndrome?

Identifiable physical characteristics of individuals with Fragile X syndrome tend to be less obvious in childhood, yet may become more so with age. If present, physical characteristics may include:

- a long, narrow face and protruding ears
- highly flexible finger joints, wrists, and elbows
- flat feet
- low muscle tone
- soft skin

Individuals with Fragile X may have delays in:

- speech and language
- motor skills such as crawling, walking, and toileting

Individuals with Fragile X syndrome often have an intellectual disability. Levels of intellectual functioning consistently tend to be lower in males and more variable in degree in females.

Fragile X syndrome also can result in problems relating to others, which can range from shyness and social anxiety to autism. Hand-flapping, hand-biting, impulsivity, poor attention span, and difficulty making eye contact/gaze avoidance may be present.

Can Fragile X syndrome be treated?

There is no cure for Fragile X syndrome. However, early intervention in key areas may be effective in improving quality of life and helping individuals with Fragile X to reach their full potential. Speech and language, occupational, and behavior therapies can be very successful. Medications may be helpful in treating anxiety, hyperactivity, and poor attention span.

In educational settings, the use of pictures, diagrams, and familiar contexts can be very useful teaching tools. Individuals with Fragile X syndrome often have strong visual memories and learn well through these methods. Individuals with Fragile X syndrome may be eligible for special education services, depending upon intellectual functioning levels.

It is not uncommon for Fragile X carriers to feel guilt for having a transmittable genetic disorder. Supportive psychotherapies and counseling may prove to be of value. These therapies may also be useful in addressing issues of shyness, depression, and worry in individuals with Fragile X.

It will be important to evaluate an individual's strengths and challenges with healthcare providers to design an individualized treatment plan. Revisit the plan often and make adjustments as necessary.

Please see reverse for resources.



Fragile X Syndrome

RESOURCES

Who We Are and Who We Serve

The **Vanderbilt Kennedy Center (VKC)** works with and for people with disabilities and their family members, service providers and advocates, researchers and policy makers. It is among only a few centers nationwide to be a University Center for Excellence in Developmental Disabilities, a *Eunice Kennedy Shriver* Intellectual and Developmental Disabilities Research Center, and a Leadership Education in Neurodevelopmental and Related Disabilities Training Program. The following are some of the ways the Center's programs and staff can assist families, educators, and other service providers.

Two Easy Ways to Take Part in Research

The Vanderbilt Kennedy Center serves families through research studies. StudyFinder is a searchable database that lists current VKC studies, including ASD research. Studies seek people of all ages with and without developmental disabilities. See kc.vanderbilt.edu/studyfinder, (615) 936-0448. Research Match is a secure place for volunteers and researchers to connect. Once you sign up and get added to the registry, a researcher will contact you if you're a possible match for the research study. See www.researchmatch.org.

Tennessee Disability Pathfinder

Tennessee Disability Pathfinder is a free statewide phone, web, and print referral service in English and Spanish. It connects the Tennessee disability community with service providers and resources. Its website database has over 3,000 agencies searchable by Tennessee county and service. Pathfinder is a project of the VKC, TN Council on Developmental Disabilities, TN Department of Health, and the TN Department of Intellectual and Developmental Disabilities. Contact www.familypathfinder.org, (615) 322-8529, toll-free (800) 640-4636.

Fragile X Clinic

This Vanderbilt Department of Pediatrics' Clinic provides resources for individuals and families affected by fragile X. The Clinic is part of the National Fragile X Foundation Clinics Consortium. It serves children and young adults who have a diagnosis of fragile X syndrome or fragile X premutation carriers. Evaluation and consultation services are available. Contact (615) 936-0249.

Reading Clinic

This clinic provides intensive, evidence-based instruction and assessment for students in kindergarten through middle school. Contact readingclinic@vumc.org or (615) 936-5123.

Sibling Supports

Support for siblings who have a brother or sister with a disability, chronic health care issue, or mental health concern. SibSaturdays, ages 5 to 12 and Tennessee Adult Brothers and Sisters (TABS), ages 18+. Contact (615) 936-8852.

Other Local and National Resources

- Fragile X Research Foundation
www.fraxa.org, (978) 462-1866
- The National Fragile X Foundation
www.fragilex.org, (800) 688-8765
- National Institute of Child Health and Human Development (NICHD)
www.nichd.nih.gov/health/topics/fragilex/Pages/default.aspx
- The Fragile X Treatment Research Program at Vanderbilt University Medical Center
(615) 936-3288
- The Arc US
www.thearc.org, (301) 565-3842
- The Arc of Tennessee
www.thearctn.org, (800) 835-7077, (615) 248-5878
- Tennessee Developmental Disabilities Network
www.tennddnetwork.org
- Tennessee's Early Intervention System (TEIS)
(800) 852-7157
- Health Library, Jr. League family Resource Center, Monroe Carrell Jr. Children's Hospital at Vanderbilt
www.vanderbiltchildrens.com/healthlibrary

Contact the Vanderbilt Kennedy Center Nashville (615) 322-8240

Toll-Free (866) 936-VUKC [8852]

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