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.

VANDERBILT KENNEDY CENTER
FOR EXCELLENCE IN DEVELOPMENTAL DISABILITIES

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, educators and service providers, researchers, students, and policy makers. Faculty and staff engage in interdisciplinary research, training, service, and information dissemination and work in collaboration with local, state and national networks and partners.

Take Part in Research

StudyFinder is a searchable database that lists current VKC studies. Studies seek people of all ages with and without developmental disabilities. See vkc.vumc.org/studyfinder. **Research Match** is a secure place for volunteers and researchers to connect. See researchmatch.org

Tennessee Disability Pathfinder

Tennessee Disability Pathfinder provides free information, referral sources, and help with navigating services via phone, email, and website. Assistance is available to individuals of all ages, all types of disabilities, and all languages spoken. Its website database has more than 3,000 agencies searchable by Tennessee county and service. Pathfinder is a project of the VKC and is partially funded by Tennessee Council on Developmental Disabilities and other state agencies. (615) 322-8529, toll-free (800) 640-4636, TNPathfinder.org

Reading Clinic

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

Sibling Supports

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

Other Local and National Resources

- Fragile X Research Foundation
  www.fraxa.org
- The National Fragile X Foundation
  www.fragilex.org
- National Institute of Child Health and Human Development (NICHD)
  www.nichd.nih.gov/health/topics/fragilex/Pages/default.aspx
- The Center for Child Development at Monroe Carell Jr. Children's Hospital at Vanderbilt
  (615) 936-0249
- The Arc US
  www.thearc.org
- The Arc of Tennessee
  www.thearctn.org
- Tennessee Developmental Disabilities Network
  www.tennddnetwork.org
- Tennessee's Early Intervention System (TEIS)
  tn.gov/didd/teis
- Junior League Family Resource Center,
  Monroe Carrell Jr. Children's Hospital at Vanderbilt
  www.childrenshospitalvanderbilt.org/information/family-resource-center

Contact the Vanderbilt Kennedy Center

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