

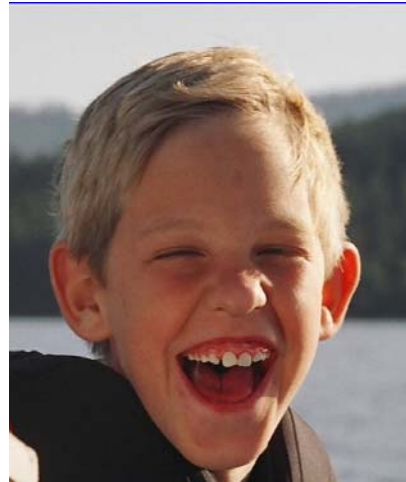
# Fragile X Syndrome

## TIPS AND RESOURCES

### 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 information about Vanderbilt Kennedy Center programs.

# Fragile X Syndrome

## TIPS AND RESOURCES

### Vanderbilt 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) 936-0282, [jennifer.hamilton@vanderbilt.edu](mailto:jennifer.hamilton@vanderbilt.edu).

### Who We Are and Who We Serve

The Vanderbilt Kennedy Center works with and for people with disabilities and family members, service providers and advocates, researchers and policy makers. It is among only a few centers nationwide to be both a University Center for Excellence in Developmental Disabilities, Education, Research, and Service and a national *Eunice Kennedy Shriver* Intellectual and Developmental Disabilities Research Center.

### Two Ways to Take Part in Research

The Vanderbilt Kennedy Center serves families of people with Fragile X syndrome through research studies.

**StudyFinder** is a searchable database that lists current studies. Studies seek people of all ages with and without developmental disabilities. See [www.kc.vanderbilt.edu/studyfinder](http://www.kc.vanderbilt.edu/studyfinder), (615) 936-0448.

**Research Family Partners** allows families with and without disabilities to register with the Center and to be contacted when a study begins for which the family might be eligible. See [www.kc.vanderbilt.edu/rfp](http://www.kc.vanderbilt.edu/rfp), (615) 936-0448.

### Tennessee Disability Pathfinder

This resource and referral service provides free phone, web, and print resources in English and Spanish. It is a project of the Vanderbilt Kennedy Center and the Tennessee Council on Developmental Disabilities. See [www.familypathfinder.org](http://www.familypathfinder.org), (615) 322-8529, (800) 640-4636.

### Family Outreach Center

This Center provides families with a single point of entry into the many services and supports of the Vanderbilt Kennedy Center, Vanderbilt University, and the community, including research projects, clinics, social work services, and recreational and arts programs. Contact [familyoutreach@vanderbilt.edu](mailto:familyoutreach@vanderbilt.edu), (615) 936-5118.

### Behavior Analysis Clinic

This clinic provides in-depth assessments of the causes of behavioral problems and develops family oriented person-centered interventions. The clinic serves children ages 5 and up. Contact [n.houchins-juarez@vanderbilt.edu](mailto:n.houchins-juarez@vanderbilt.edu), (615) 322-9007.

### Reading Clinic

This clinic provides intensive, individualized, one-to-one tutoring using assessment and instructional methods proven by research to promote reading. The clinic serves children in early elementary grades. Contact [caresa.l.young@vanderbilt.edu](mailto:caresa.l.young@vanderbilt.edu), (615) 936-5123.

### Sibling Supports

Sibling programs are provided for siblings who have a brother or sister with a disability. Groups for children, teens, and adults give siblings the opportunity to meet other siblings, and to participate in age-appropriate group discussions and activities. Contact (615) 936-5188, [familyoutreach@vanderbilt.edu](mailto:familyoutreach@vanderbilt.edu).

### Other Local and National Resources

#### Fragile X Research Foundation

[www.fraxa.org](http://www.fraxa.org), (978) 462-1866

#### The National Fragile X Foundation

[www.fragilex.org](http://www.fragilex.org), (800) 688-8765

#### National Institute of Child Health and Human Development (NICHD)

[www.nichd.nih.gov/health/topics/fragile\\_x\\_syndrome.cfm](http://www.nichd.nih.gov/health/topics/fragile_x_syndrome.cfm)

#### The Arc US

[www.thearc.org](http://www.thearc.org), (301) 565-3842

#### The Arc of Tennessee

[www.thearctn.org](http://www.thearctn.org), (800) 835-7077, (615) 248-5878

#### Tennessee Developmental Disabilities Network

[www.tennddnetwork.org](http://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](http://www.vanderbiltchildrens.com/healthlibrary)

#### Contact the Vanderbilt Kennedy Center

Nashville (615) 322-8240

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

[kc@vanderbilt.edu](mailto:kc@vanderbilt.edu)

