Fragile X Syndrome

Fragile X syndrome is the most common inherited cause of mental retardation in boys. It is usually caused by a gene abnormality passed on from mother to son. Certain physical abnormalities and behavior problems are also present. There is no cure for fragile X syndrome. Special education and various types of therapy can be helpful for children with fragile X syndrome and their families.

What is fragile X syndrome?

Fragile X syndrome is a relatively common genetic disease causing mental retardation. It is more common and severe in boys, although it can occur in girls. The disease is most often caused by an abnormal gene passed on from mothers to sons. Fragile X syndrome does not result from anything the parents did wrong or anything that happened during pregnancy or childbirth.

Most children with fragile X syndrome have problems with intellectual functioning. Some have relatively mild learning disabilities; others have moderate mental retardation. Some physical abnormalities occur along with certain medical problems. Social and behavior problems are common as well.

There is no cure for fragile X syndrome. However, special education and other services can help to make the most of your child's intellectual abilities and other skills. The benefits are greatest if the condition is recognized and services are started as early as possible. Genetic counseling can help you to understand your risk of passing the disease on to future children.

What kinds of problems occur with fragile X syndrome?

Intelligence/development.

- Children with fragile X syndrome have lower than normal intelligence. Your child may be slow to reach normal language milestones. In less severe cases, the problem may not be detected until your child reaches school age. In girls with the syndrome, intelligence may be closer to normal.
- Language and speech problems, such as difficulty using the right words and problems with pronunciation (such as stuttering).
- Emotional and behavioral difficulties, especially in boys. Your child may be shy and anxious, especially around unfamiliar people and situations. In these circumstances, angry outbursts or temper tantrums may occur. Sometimes it may be difficult to tell what's making your child upset.
- Children with fragile X syndrome may be diagnosed with other psychiatric or developmental disorders, especially attention deficit–hyperactivity disorder and autism.

Physical abnormalities.

- In infants and young children, appearance is normal. During puberty, your child develops a large face, jaw, and ears. In boys, the testicles may be enlarged (sexual function is normal).
- Loose joints ("double-jointed") are related to an abnormality of the body's connective tissue. This can lead to problems such as joint dislocations or hernias.

Medical problems.

- Heart problems. Later in life, the connective tissue abnormalities may lead to heart problems such as mitral valve prolapse.
- Girls may have premature ovarian failure, causing early menopause. This means the ovaries are no longer producing eggs and therefore pregnancy cannot occur.

Individual symptoms and behaviors vary a lot. Your child may have different symptoms or the problems may be more or less severe.

What causes fragile X syndrome?

- This disease is caused by an abnormality in the fragile X mental metardation 1 (*FMR1)* gene. The *FMR1* gene makes proteins that affect how the brain works.
- The *FMR1* gene mutation is found on the X chromosome. Females have two X chromosomes (XX), while males have an X chromosome and a Y chromosome (XY). Since females have two X's, one of them will be normal; that's why girls are less likely than boys to be affected by fragile X syndrome. Males can only pass the abnormal *FMR1* gene to daughters, not sons. This is because males have to donate a Y chromosome (not an X) to have a male child.

How is fragile X syndrome diagnosed?

- Fragile X syndrome is recognized based on your child's symptoms, a physical examination, and information on your family history. Once the diagnosis is suspected, it can be confirmed by a special chromosome test.
- Genetic counseling can help you to understand the risks of having another child with fragile X syndrome.

How is fragile X syndrome managed?

Many of the problems associated with fragile X syndrome can be improved with treatment. It is especially important to seek help with behavioral or emotional disorders, which can be stressful for your family.

- Education is probably the most important aspect of care:
  - *Early intervention* should start as soon as your child's condition is diagnosed. Every state has an early intervention program; our office can put you in touch with resources to get you started. Early intervention experts can assess your child and develop an Individualized Family Support Plan (IFSP) based on your child's development, need for support, and goals for independence.
  - *Special education* services are also available in every state (although children with less severe impairment may not qualify for services). You are entitled to expert evaluation of your child. Based on the results, an Individualized Education Program (IEP) can be developed to meet your child's educational needs.
- Several professionals may play a role in your child's care. For example:
  - A speech/language pathologist can help with stuttering or other language difficulties.
  - An occupational therapist may help in identifying the best adaptations to your child's needs.
  - A behavioral therapist may help in dealing with emotional and behavioral problems.
  - Other types of services can be helpful. Family counseling may help to deal with the emotional or social problems associated with having a special needs child.

When should I call your office?

Call our office if you have any questions about your child's testing, treatment, or educational intervention.

Where can I get more information about fragile X syndrome?

- Contact the National Fragile X Foundation on the Internet at *www.fragilex.org*, or call 1-800-688-8765.
- The National Institute of Child Health and Human Development (NICHD) offers an informative booklet on "Families and Fragile X Syndrome." It is available on the Internet at *www.nichd.nih.gov/publications/pubs/fragileX/*, or call 1-800-370-2943.

Please type your custom instructions and/or office contact information here.