



FROM
YOUR DOCTOR

Fragile X syndrome and your child

Fragile X syndrome is a genetic disorder caused by a mutation (a change in the DNA structure) in a gene called the fragile X mental retardation 1 gene, or *FMR-1*. It is the most common known cause of inherited mental retardation. You or other members of your family did not do anything to cause this syndrome. The gene mutation usually is carried unknowingly by families for many generations. Your child may be the first one in your family to show signs of fragile X syndrome, or you may have other relatives who are affected.

How does fragile X syndrome affect children?

Fragile X syndrome can cause hyperactivity, learning or emotional problems in children with a normal IQ, and any level of mental retardation. Boys are usually affected more severely than girls. Most are mentally retarded, meaning that their IQ is below 70. In contrast, only about 30% of girls with fragile X syndrome are mentally retarded, though many have learning problems and a short attention span. Carriers of the fragile X gene mutation can be completely normal. (Carriers have a small change in the *FMR-1* gene, whereas children affected by fragile X syndrome have a large change in the *FMR-1* gene.)

• **Physical features** Boys with fragile X syndrome often have prominent ears, a large head, a prominent forehead, extra-flexible finger joints, a high palate, soft, velvety skin, and flat feet. During puberty, they usually develop large testicles and a somewhat long face. Many of these features occur in the general population, so most patients with physical features of fragile X syndrome look normal. Girls may also have physical features associated with fragile X syndrome, particularly large or prominent ears, very flexible finger joints, and flat feet. In general, children with fragile X syndrome do not develop medical complications, although they may have frequent ear infections and are at higher risk than the average child for seizures, hernia, and joint dislocation.

• **Behavior and development** The major problems associated with fragile X syndrome involve behavior, slow development, and a lower IQ. Children with the syndrome tend to be extra sensitive to stimuli in their environment such as noises or changes in their routine. They are often anxious and may have frequent tantrums. Girls especially are prone to shyness, social anxiety, and moodiness. Boys and some girls may have high activity levels and short attention spans. Language development is especially slow, so that most boys do not talk much by 3 years of age. Children with fragile X syndrome commonly develop unusual behaviors by age 3, including hand flapping, hand biting, poor eye contact, excessive chewing on clothes, and pulling away from touch. They may ask questions over and over again even after hearing the answer.

• **Strengths** Children with fragile X syndrome also have many strengths. These include their social abilities (although shyness is a common trait), a wonderful sense of humor, an excellent memory for people, events, and directions, intense interests, and outstanding imitation skills.

How is the syndrome diagnosed?

Your doctor or a genetic counselor will review how the gene mutation is passed on in your family and who should be tested for it. There are two types of blood test for fragile X syn-

Fragile X syndrome and your child

continued

drome. One is called a chromosome or cytogenetic test. The other is a newer DNA test that shows the change, or mutation, in the *FMR-1* gene structure. This mutation "turns off" the *FMR-1* gene and causes the problems associated with fragile X syndrome. Once the diagnosis is made, fragile X syndrome can be prevented in future pregnancies by means of such new reproductive technologies as in vitro fertilization and egg donation. Your genetic counselor can discuss prenatal diagnosis and prevention with you.

What can be done to treat fragile X syndrome?

Although there is no cure for fragile X syndrome, many treatments are available. If your child is diagnosed in infancy, your doctor will discuss infant stimulation programs and developmental preschool programs with you as your child grows. Speech and language therapy, occupational therapy, and physical therapy are important to aid your child's motor and language development. A type of occupational therapy called sensory integration can help with behavioral problems. The occupational therapist will demonstrate physical techniques, such as deep pressure back rubs and joint compression, that may help calm behavior outbursts or tantrums at home. Your doctor or psychologist can help you establish a program to control behavior at home that includes consistent structure, regular routines, and positive feedback.

In school, your child's special education program should include participation in regular classes whenever possible, individualized help to modify assignments and tutor weak academic areas, speech and language therapy, occupational therapy, and use of computers to teach language and academic skills. Your doctor can help

you advocate for these services at your child's school and obtain appropriate therapies when needed.

Your doctor will treat any medical problems that may accompany fragile X syndrome. They include seizures in 20% of children and strabismus (a weak eye muscle) in 30%. Mitral valve prolapse, a heart condition, occurs in 50% of adults. When necessary, your physician may prescribe medications to improve poor attention span, hyperactivity, tantrums, aggression, anxiety, obsessive thinking, or other behavioral or emotional difficulties. These medications can work together with other treatments to improve your child's learning and behavior. Your doctor will explain the side effects of any recommended medication. Medications are given for a trial period and should be continued only if they prove helpful. If your child has an adverse response to medication, call your doctor to discuss it.

Where to find help

When fragile X syndrome is diagnosed, it often helps to talk to another family with a child who is similarly affected. The National Fragile X Foundation has a list of resource centers and parent support groups throughout the United States. You can phone the foundation at (800) 688-8765 or (303) 333-6155 for educational information and the phone number of a parent near you who has a child with fragile X syndrome.

Children with fragile X syndrome are difficult to raise, and parents need personal time and recovery time on a regular basis. If you feel overwhelmed, depressed, or excessively anxious, discuss this with your doctor. He or she can help you. When you are healthy, you can better help your child reach his or her greatest potential.