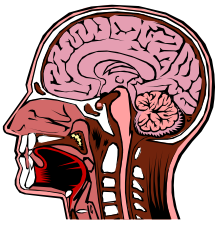
Disability Awareness Begins With You



Fragile X Syndrome

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What is fragile X syndrome?

Fragile X syndrome is an inherited genetic condition associated with mental retardation. It is identified by a break, or weakness, on the long arm of the X chromosome. This is an abnormality of a sex chromosome. The term "sex-linked" or "X-linked" inheritance is used in the medical literature. This means that mothers are carriers and their sons are at risk of being affected, while daughters are at risk of being carriers and sometimes mildly affected.

How common is fragile X syndrome?

The prevalence of carriers in the general population is approximately 1 in 600 (Sherman, 1992). If a 3 percent prevalence of mental retardation in the general population is used, fragile X syndrome may account for up to 10 percent of mental retardation. It is the most common inherited cause of mental retardation known to exist.

Why aren't girls affected as much as boys?

The X chromosome is unique among the 23 pairs of chromosomes found in each human cell. It is one-half of the chromosome pair that determines sex. Males can only inherit a fragile X from their mothers. Disorders which are carried on the X chromosome usually demonstrate less involvement in females because a second X chromosome can compensate for a defective gene on the other X chromosome. In 30 percent of females who inherit the fragile X gene will be affected intellectually by this syndrome, however, more commonly demonstrate learning disabilities including math deficits and problems with attention span even though their IQ may be within the normal range.

What are the physical characteristics of fragile X syndrome?

The physical features associated with fragile X syndrome include a long narrow face, prominent ears, jaw and forehead. Enlarged testicles, known as macroorchidism, is another physical characteristic. Many young children with fragile X syndrome may not show these features although prominent ears are seen in approximately two-thirds of children. Loose joints, particularly in the finger joints, are also quite common in childhood. After puberty these features are more common, particularly the long face and macoorchidism. Physical features are often more subtle in females, but approximately 50 percent of females experience them.

What degree of mental retardation is associated with fragile X syndrome?

About 80 percent of boys who inherit the fragile X have mental impairment, ranging from severe retardation to low-normal intelligence. The majority are mildly to moderately retarded. Girls are much less affected, with estimates that about 30 percent with the genetic condition have some degree of mental retardation (Sherman, et al, 1985). Recent research suggests that IQs of males with the fragile X syndrome appear to decline throughout childhood. While young boys may be only mildly impaired, adults with the fragile X syndrome are usually moderately or severely retarded. This is because their rate of learning does not keep pace with their initial IQs as they grow older (Lachiewicz, et al, 1987).

What behavioral symptoms are associated with fragile X?

Men and boys with the fragile X syndrome are usually socially engaging, but they have an unusual style of interacting with other people (Meryash, 1985). They tend to avoid direct eye contact during conversation, and hand-flapping or hand-biting is common. They may have an unusual speech pattern characterized by a fast and fluctuating rate and repetitions of sounds, words or phrases. They also may have a problem in their attention span, hyperactivity, and motor delays. Some males demonstrate autistic-like behaviors, including perseverative speech, unusual hand mannerisms and problems in relating to others (Hagerman, 1991).

Can fragile X be treated?

There is no cure for fragile X syndrome, but medical intervention can improve the problems in attention and hyperactivity of many young boys (Hagerman, 1991; Hagerman and McKenzie, 1992). A variety of medications can improve attention span, concentration, hyperactivity, aggressive behavior and other problems (Hagerman, 1991).

Treatment for children with fragile X syndrome is determined by an individualized program of special education including such components as speech therapy, physical therapy and vocational preparation. Children who are living at home and enrolled in special education will function at a higher level as adults than do men who were institutionalized years ago and never received special education (Meryash, 1985). Early intervention and preschool education become especially important, for evidence indicates that while the rate of learning slows with age, males with fragile X syndrome do not lose previously acquired skills or deteriorate in functioning (Lachiewicz et al, 1987).

When was fragile X syndrome identified?

Although it was well-known for years that more males than females are affected by mental retardation, it was the 1960s before scientists perfected ways to look at chromosomes and detect abnormalities. The fragile site was first noted in 1969 by Dr. Herbert Lubs who discovered these chromosomes in a family having two brothers with mental retardation. By the late 1970s this condition was named fragile X. and was published in the medical press . It has only come to the attention of the public in the last ten years. Because of its recent discovery, most individuals with this syndrome are undiagnosed. DNA testing is now available for identifying both carriers and individuals affected by the fragile X syndrome (Verkerk, et al, 1991).

What are the chances of having a child with fragile X?

The DNA testing has demonstrated that all individuals who demonstrate the fragile X syndrome have a parent who is a carrier. If the person affected by fragile X is a male, the carrier is always his mother. Since the carrier mother has two X chromosomes the chance of passing on the fragile X chromosome is 50 percent. Since the inheritance pattern in fragile X is complex, it is important for family members to receive genetic counseling concerning their risks of carrying the fragile X gene and passing it on to their children.

Where can information about genetic counseling be obtained?

The National Center for Education in Maternal and Child Health (NCEMCH) is a major link between sources of information, services and the professional in areas of maternal and child health, including medical genetics. NCEMCH's address is 3520 Prospect St., N.W., Washington, D.C. 20057. The National Fragile X Foundation also exists with a network of over 50 resource centers throughout the United States and internationally. Its address is 1441 York St., Suite 215, Denver, Colorado 80206, and the telephone number is 1-800-688-8765.