

Fragile X

Definition

Fragile X syndrome, also called Martin-Bell syndrome, is an inherited genetic condition often associated with intellectual disability.

It is identified by a break, or weakness, on the long arm of the X chromosome. Mothers are carriers and their sons are at risk of being affected, while daughters are at risk of being carriers and sometimes mildly affected. While more boys than girls are affected by fragile X, it is not transmitted from father to son.

Because of its recent discovery, most individuals with this syndrome are undiagnosed.

Characteristics

- May have high arched palate, long narrow face, prominent ears, jaw and forehead. Loose joints, particularly in the finger joints.
- About 80 percent of boys who inherit the fragile X also have intellectual disability.
- Socially engaging, but have an unusual style of interacting with other people.
- Tend to avoid direct eye contact during conversation, and hand-flapping or hand-biting is common.
- May have an unusual speech pattern characterised by a fast and fluctuating rate and repetitions of sounds, words or phrases.
- Also may have a problem in attention span, hyperactivity, and motor delays.
- Some males demonstrate autistic-like behaviours, including perseverative speech, unusual hand mannerisms and problems in relating to others.

Diagnosis

DNA testing is now available to identify unaffected carriers and individuals who are affected by the fragile X syndrome.

Prevalence

Believed to be 1 in 1,000. Is the most common mutation in the human genome that causes intellectual difficulties. Boys are more affected.

Prognosis

There is no cure for fragile X syndrome, but medical intervention can improve the problems in attention and hyperactivity.

Strategies

- Currently gene therapy and gene repair are being explored. Some currently available medications can relieve symptoms of fragile X.
- Need individualized program of special education including such components as:
 - learning program
 - speech therapy,
 - physical therapy
 - vocational preparation.
- Early intervention and preschool education are especially important.