

Fragile X Syndrome

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Fragile X syndrome, called Martin-Bell syndrome, is a genetic disorder and is the most common form of inherited mental retardation. It is a sex-linked genetic abnormality in which a mother is a carrier, transmitting the disorder to her sons. It affects approximately 1 in every 1,000 to 2,000 male individuals, and the female carrier frequency may be substantially higher. Males afflicted with this syndrome typically have a moderate to severe form of intellectual handicap. Females may also be affected but generally have a mild form of impairment.

Approximately 15% to 20% of those with Fragile X Syndrome exhibit autistic-type behaviors, such as poor eye contact, hand-flapping or odd gesture movements, hand-biting, and poor sensory skills. Behavior problems and speech/language delay are common features of Fragile X Syndrome.

People with Fragile X syndrome also have a number of recognizable physical features, including a high arched palate, strabismus (lazy eye), large ears, long face, large testicles in males, poor muscle tone, flat feet, and sometimes mild, heart valve abnormalities. Although most individuals with Fragile X syndrome have a characteristic 'look' (long face and large ears), there are some who do not have typical features.

Many hospitals and laboratories perform blood tests to diagnose Fragile X syndrome. Several treatments are recommended for individuals with this disorder, including mild medications for behavior problems and therapies for speech and language and sensory improvement. Also, families are advised to seek genetic counseling to understand the inheritable nature of Fragile X Syndrome and to discuss with family members the likelihood other individuals or future offspring may have this disorder.