

Fragile X syndrome is characterized by moderate intellectual disability in affected males and mild intellectual disability in affected females. Distinctive physical features are sometimes present in affected males including a large head, long face, prominent forehead and chin, protruding ears, loose joints and large testes, but these features develop over time and may not be obvious until puberty. Motor and language delays are usually present but also become more apparent over time. Behavioral abnormalities including autistic behaviors are common. Fragile X syndrome is characterized by moderate intellectual disability in affected males and mild intellectual disability in affected females. The physical features in affected males are variable and may not be obvious until puberty. These symptoms can include a large head, long face, prominent forehead and chin, protruding ears, loose joints and large testes. Other symptoms can include flat feet, frequent ear infections, low muscle tone, a long narrow face, high arched palate, dental problems, crossed eyes (strabismus) and heart problems including mitral valve prolapse. Delayed motor development, hyperactivity, behavior problems, toe walking, and/or occasional seizures can also occur in some patients. Autistic behaviors such as poor eye contact, hand flapping, and/or self-stimulating behaviors are also common. Motor and language delays are usually present but become more apparent over time. The fragile X syndrome affects about 1 in 4,000 males and 1 in 6,000 to 8,000 females in the USA; that is, it affects about twice as many males as it does females. However, about four times as many females appear to be carriers of the altered gene as do males (1:250 females and 1:1000 males). Fragile X syndrome has been found in all major ethnic groups and races.