

Fragile X Syndrome

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Fragile X Syndrome (*Martin-Bell Syndrome*) is a **genetic disorder** that is caused by a break or weakness on the **X chromosome** (females have two X chromosomes and males have one X and one Y). Because of this defect, the gene cannot manufacture the protein that it normally makes, which causes the features and characteristics of the syndrome. Fragile X is estimated to occur in **1 in 1,200 males** and **1 in 2,500 females**. It occurs in all racial, ethnic, and socioeconomic groups. It is said to be one of the most common genetic diseases and is the most common inherited cause of learning disabilities and mental retardation known to exist.

Fathers who are carriers can only pass the affected gene on to their daughters. The reason for this is that for males, the father always gives a Y chromosome and the mother always gives an X chromosome (if the father gives an X chromosome instead of a Y, the child would be female). Therefore, in order for the father to pass the fragile X gene, the recipient would have to be a female. Mothers who are carriers have a 50% chance of passing on the fragile X mutation to their children. **Females tend to be more mildly affected by the fragile X gene than males** because females have a second X chromosome to compensate for the defective gene on the affected X chromosome. It is estimated that 1 in 259 females, in the general population, can carry the fragile X mutation either as premutation or full mutation. Those with premutations are generally unaffected intellectually and of those who carry the full mutation, approximately 50% will display a learning disability, intellectual deficit, or mental retardation.

Features usually but not always include:

- mental impairment, ranging from learning disabilities to mental retardation
- attention deficit and hyperactivity
- anxiety and unstable mood
- autistic behaviors
- long face, large ears, flat feet
- hyper extensible joints, especially fingers
- Seizures (epilepsy) affect about 25% of people with fragile X



Fragile X is a family of genetic conditions, which can impact individuals and families in various ways. These genetic conditions are related in that they are all caused by gene changes in the same gene, called the FMR1 gene.

Expert in Fragile X Syndrome? DSES is always looking for volunteers to help teach or give presentations!!

Resources and Additional Information

National Fragile X Foundation; www.fragilex.org

Fragile X Research Foundation; www.fraxa.org