



FRAGILE X TREME BULL BLOW OUT

RIDING FOR A REASON

What is fragile x syndrome?

Fragile X syndrome is the most common inherited cause of mental impairment. The syndrome occurs in approximately 1 in 3600 males and 1 in 4000 to 6000 females.

Impact on males: The majority of males with fragile X syndrome will have a significant **intellectual disability**. The spectrum ranges from learning disabilities to severe mental impairment and autism. In addition, males have a **variety of physical and behavioral characteristics**. However, no male has all of these characteristics.

Physical features such as enlarged ears, long face with prominent chin. **Connective tissue** problems may include ear infections, mitral valve prolapse, flat feet, double-jointed fingers, hyperflexible joints and a variety of skeletal problems.

Behavioral characteristics in males include attention deficit disorders, speech disturbances, hand biting, hand flapping, autistic behaviors, poor eye contact, and unusual responses to various touch, auditory or visual stimuli.

Impact on females: The characteristics seen in males can also be seen in females, though females often have milder intellectual disability and a milder presentation of the behavioral or physical features.

About a third of the females have a **significant intellectual disability**. Others may have more moderate or mild learning difficulties. Similarly, the physical and behavioral characteristics are often expressed to a lesser degree.

Cause A change or mutation in a gene on the X chromosome causes the fragile X syndrome. Chromosomes are packages of genes that are passed from generation to generation. Most individuals have 46 chromosomes, two of which are sex chromosomes. In females, these are two X's; in males they are one X and one Y. Genes are given names to identify them and the gene responsible for fragile X syndrome is called the **FMR1** (fragile X mental retardation 1) gene. The mutation is in the **DNA** (the chemical that makes up genes), of the X chromosome. The gene appears in three forms that are defined by the number of repeats of a pattern of DNA called CGG repeats. Individuals with less than 60 CGG repeats have a normal gene. Individuals with 60-200 CGG repeats have a premutation which means they carry an unstable mutation which can expand in future generations. Individuals with over 200 repeats have a full mutation which causes fragile X syndrome. The full mutation causes the gene to shut down or **methylyate** a region of the FMR-1 gene. Normally, the FMR-1 gene produces an important protein called FMRP. When the gene is turned off, the individual does not make fragile X mental retardation protein (**FMRP**). The lack of this specific protein causes fragile X syndrome.

Treatment: **At this time, there is no cure for fragile X syndrome.**