Living with Fragile X Syndrome

Fragile X syndrome is the most common form of inherited intellectual and developmental disability.

1. Introduction

Fragile X syndrome is an inherited genetic disorder that causes a range of learning, behavioral, and developmental challenges. Usually, males are more severely affected by this condition, with most males and about half of females with fragile X syndrome having characteristic physical features that become more apparent with age. Fragile X syndrome is also the leading known genetic cause of autism. Children with fragile X usually have delayed development of speech and language by age 2, and they may also develop anxiety and hyperactive behavior.

2. Causes

Fragile X syndrome is caused by a genetic mutation of the FMR1 gene on the X chromosome. Abnormal mutations in a DNA segment silence the FMR1 gene, leading to a shortage of the FMRP protein and disrupting nervous system function, which causes the signs and symptoms of this condition. Females can be carriers without having symptoms, but males with fragile X will always have symptoms.

3. Symptoms

The most common signs and symptoms of fragile X syndrome include intellectual disability, behavioral and learning challenges, and various physical characteristics. Specific symptoms include:

+ Attention deficit and hyperactivity, especially in young children
+ Anxiety and unstable mood
+ Autistic behaviors such as hand-flapping and not making eye contact
+ Sensory integration problems
+ Speech delay and delayed early milestones

Physical signs of fragile X syndrome may be hard to recognize in babies and young children, but they sometimes become more apparent with age:

+ Long face, prominent ears, flat feet
+ Very flexible or double-jointed fingers
+ Low muscle tone
+ A high-arched palate

4. Treatments

Fragile X syndrome does not have a cure, but you can treat its symptoms. The most common treatments include a combination of medications and therapy to help with coping and behavioral skills.

Did you know?

Researchers estimate that, worldwide, about 1 in 11,000 females and 1 in 7,000 males have fragile X syndrome.

For more information on fragile X syndrome and supportive resources, please visit fraxa.org.

References:

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