Fragile X syndrome

Fragile X syndrome is a genetic disorder that leads to intellectual disability, developmental delays, and behavioral challenges. It is caused by a mutation in the FMR1 gene (Fragile X Mental Retardation 1) located on the X chromosome. Normally, this gene produces a protein essential for brain development, but in people with Fragile X, an abnormal expansion of DNA repeats (CGG triplets) silences the gene, reducing the production of this protein.

Key Features: Intellectual disability: Typically ranging from mild to moderate, though some individuals with Fragile X syndrome have normal intelligence. Developmental delays: Children may learn to walk or talk later than usual. Behavioral challenges: Issues such as attention deficits, hyperactivity, anxiety, repetitive behaviors, and sensory sensitivities are common. Physical characteristics: Some individuals may have distinctive facial features, including a long face, large prominent ears, and a prominent jaw, although these traits may not always be present. Fragile X syndrome is more common in males because they have only one X chromosome, so if the FMR1 gene is mutated, there is no second X chromosome to compensate. Females can also be affected but usually have milder symptoms.

Treatment: While there is no cure, treatments include behavioral therapy, educational interventions, and, in some cases, medications to manage symptoms such as anxiety, hyperactivity, and attention problems.

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