



Empowering Children. Supporting Families. Building Brighter Futures.

Resource 065 | Genetic and chromosomal conditions

Fragile X syndrome

What it means

Fragile X syndrome is a genetic condition that can affect learning, language, attention, sensory regulation, and social participation. Its effects vary, including between children of different sexes. Some children also have autism or seizures. A genetic diagnosis guides counseling and care without replacing individual developmental assessment. [1]

Causes and risk factors

Fragile X syndrome usually involves a large expansion of a repeated DNA sequence in the FMR1 gene, reducing an important protein involved in brain development. Smaller expansions have different implications. The inheritance pattern is complex, so genetic counseling helps families understand testing and possible implications for relatives. [1,2]

General family education. A concern deserves assessment, but this guide does not diagnose a child or prescribe treatment.



Signs and everyday impact

Speech and language delay, learning difficulties, attention or anxiety concerns, sensory sensitivities, and differences in social interaction may occur. Physical features may become more apparent with age and are not sufficient for diagnosis. Intellectual functioning varies, and a child's communication or behavior should be interpreted in context. [1,2]

Assessment and diagnosis

Diagnosis requires appropriate FMR1 repeat-expansion testing; standard sequencing alone may not detect the relevant change. Clinicians also assess hearing, vision, development, communication, and co-occurring conditions. Genetic counseling explains the result and distinguishes a full mutation from a premutation or other finding. [1,2]

Treatment and therapy

Individualized educational support, speech-language therapy, occupational therapy, and communication aids address functional needs. Predictable routines and developmentally appropriate psychological support can help anxiety and participation. Medicines may treat selected co-occurring problems such as seizures or ADHD under clinical supervision. There is no single established treatment that resolves all features of the syndrome. [1,2,3]

Support at home

Prepare the child for transitions with pictures, simple explanations, and time to adjust. Reduce overwhelming sensory demands when helpful. Use the child's preferred communication methods and meaningful interests. Practice one everyday skill in small steps, and coordinate supports across home, childcare, and school.

When to seek help

Seek prompt review for skill loss, possible seizures, marked behavioral change, or persistent sleep and feeding difficulties. Use emergency services for prolonged seizures, breathing difficulty, or immediate danger.

Questions for your care team

- Was the correct FMR1 expansion test performed?
- What does this result mean for family counseling?
- Which communication, learning, or anxiety supports fit this child?



Podcast preparation

Use this outline to develop a later podcast script.

Suggested opening

Welcome to Mariel Early Intervention. Today we discuss fragile x syndrome. Fragile X syndrome is a genetic condition that can affect learning, language, attention, sensory regulation, and social participation. This is general education; each child needs individualized care.

Opening: Define the topic and its developmental context.

Understanding: Explain causes and signs without inviting self-diagnosis.

Assessment: Describe evaluation and distinguish screening from diagnosis.

Support: Discuss treatment, therapy, and practical family supports.

Closing: Explain urgent signs and one clear next step.

Misconception to address: Myth: appearance alone can identify fragile X syndrome. Appropriate genetic testing is needed.

Before recording: update clinical references and local service details, arrange clinical review, and use fully consented or fictional examples.

Terms in plain language

- FMR1: the gene most often involved in fragile X syndrome.
- Repeat expansion: an unusually large number of repeated DNA units.
- Genetic counseling: support to understand genetic findings and family implications.

Sources and further reading

Sources accessed 3 October 2026. Numbered references support the clinical overview. Treatment availability and referral routes vary by country.

[1] Fragile X syndrome: MedlinePlus Genetics

<https://medlineplus.gov/genetics/condition/fragile-x-syndrome/>

[2] What are the treatments for Fragile X syndrome?

<https://www.nichd.nih.gov/health/topics/fragilex/conditioninfo/treatments>

[3] Assessing Developmental Delays in Children

<https://www.healthychildren.org/English/ages-stages/toddler/Pages/Assessing-Developmental-Delays.aspx>