



Empowering Children. Supporting Families. Building Brighter Futures.

Resource 067 | Genetic and chromosomal conditions

Angelman syndrome

What it means

Angelman syndrome is a genetic condition affecting development, communication, movement, and often sleep and seizures. Spoken language is usually substantially limited, but communication can develop through other methods. Support should recognize the child's interests and emotional needs rather than relying on stereotypes about personality. [1]

Causes and risk factors

Angelman syndrome results from loss of effective function of the maternally inherited UBE3A gene in the brain. Several genetic mechanisms can cause this, including a chromosome deletion or gene change. The mechanism affects family counseling, so genetic results require specialist interpretation. [1,2]

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



Signs and everyday impact

Developmental delay, limited spoken language, balance or movement difficulties, sleep disturbance, and seizures may occur. Some children have frequent smiling or laughter, but they also experience discomfort, frustration, and a full range of emotions. Communication needs and abilities vary and should be assessed directly. [1,2]

Assessment and diagnosis

Clinical assessment reviews development, movement, seizures, and associated features. Genetic testing may include DNA methylation analysis and additional tests to identify the underlying mechanism. A negative initial test may require further specialist evaluation. Hearing, vision, nutrition, mobility, sleep, and communication assessments guide support. [1,2]

Treatment and therapy

Care commonly includes seizure treatment, sleep management, physical and occupational therapy, and speech-language therapy focused on accessible communication. AAC can include signs, pictures, or devices and should be introduced according to need. Feeding, orthopedic, and other medical concerns receive targeted care. Developmental and educational supports should be coordinated across settings. [1,2,3]

Support at home

Make AAC available throughout the day, including for saying no, asking for help, and expressing discomfort. Use predictable routines and supported choices. Follow movement and seizure plans, and track sleep patterns for the care team. Include enjoyable play and social activities without assuming laughter always means comfort.

When to seek help

Seek urgent assessment for prolonged or changed seizures, marked reduction in alertness, or inability to maintain hydration. New skill loss or major sleep and movement changes warrant medical review.

Questions for your care team

- Which genetic mechanism caused the syndrome?
- How can we provide reliable communication access?
- What seizure, sleep, and mobility plans are needed?



Podcast preparation

Use this outline to develop a later podcast script.

Suggested opening

Welcome to Mariel Early Intervention. Today we discuss angelman syndrome. Angelman syndrome is a genetic condition affecting development, communication, movement, and often sleep and seizures. 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: a child who smiles often is always comfortable. Pain and distress still need attention.

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

Terms in plain language

- UBE3A: a gene whose brain function is important in Angelman syndrome.
- Methylation testing: a test that can identify certain chromosome-control patterns.
- Ataxia: difficulty coordinating balance and movement.

Sources and further reading

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

[1] Angelman syndrome: MedlinePlus Genetics

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

[2] Angelman Syndrome - GeneReviews® - NCBI Bookshelf

<https://www.ncbi.nlm.nih.gov/books/NBK1144/>

[3] Epilepsy and Seizures

<https://www.ninds.nih.gov/node/647>