



Empowering Children. Supporting Families. Building Brighter Futures.

Resource 069 | Genetic and chromosomal conditions

Williams syndrome

What it means

Williams syndrome is a genetic condition that can affect the heart and blood vessels, growth, development, and learning. Children may have an uneven profile of strengths and difficulties. Social or language characteristics vary, so assumptions about friendliness or ability should not replace an individual assessment. [1]

Causes and risk factors

Williams syndrome usually results from a deletion of several genes in a region of chromosome 7. Loss of the ELN gene contributes to blood-vessel and connective-tissue differences. Most cases occur as a new genetic change, although inheritance is possible. Genetic counseling explains the finding and implications. [1,2]

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



Signs and everyday impact

Possible features include feeding difficulty, slower growth, developmental delay, learning differences, anxiety, sound sensitivity, and cardiovascular problems. Elevated calcium or other endocrine concerns may occur. The combination and severity vary. A child who speaks readily may still need substantial support with understanding, motor skills, or safety. [1,2]

Assessment and diagnosis

Genetic testing, commonly a chromosomal microarray or a targeted test, confirms the deletion. A clinical team assesses cardiovascular health, growth, calcium and endocrine concerns, hearing, vision, and development. An individualized learning assessment helps identify specific strengths and needs rather than relying on a typical syndrome profile. [1,2]

Treatment and therapy

Cardiology care and ongoing surveillance are central; some children need procedures or surgery. Feeding and nutritional care, physical and occupational therapy, speech-language support, and educational accommodations address daily needs. Anxiety and sensory distress may need additional support. Before anesthesia or sedation, the medical team should review condition-specific cardiovascular risks and planning. [1,2,3]

Support at home

Use visual demonstrations and manageable steps for new tasks. Support sound comfort, predictable routines, and safe social boundaries. Follow the child's specific nutritional and activity advice; do not change calcium or vitamin intake without clinical guidance. Keep surveillance appointments and share the diagnosis with procedural teams.

When to seek help

Seek urgent care for fainting, chest pain, major breathing difficulty, or severe illness. New persistent vomiting, poor intake, or marked lethargy warrants assessment. Ensure anesthesia providers know the diagnosis before planned procedures.

Questions for your care team

- Which heart and blood-pressure follow-up is required?
- Are calcium, hearing, and endocrine checks due?
- What should procedural or anesthesia teams know?



Podcast preparation

Use this outline to develop a later podcast script.

Suggested opening

Welcome to Mariel Early Intervention. Today we discuss Williams syndrome. Williams syndrome is a genetic condition that can affect the heart and blood vessels, growth, development, and learning. 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: sociability means a child has no learning or anxiety needs. Support should follow individual assessment.

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

Terms in plain language

- Deletion: loss of a segment of genetic material.
- ELN: a gene involved in making elastin, a connective-tissue protein.
- Supravalvular aortic stenosis: narrowing above the heart's aortic valve.

Sources and further reading

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

[1] Williams syndrome: MedlinePlus Genetics

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

[2] Williams Syndrome - GeneReviews® - NCBI Bookshelf

<https://www.ncbi.nlm.nih.gov/books/NBK1249/>

[3] Assessing Developmental Delays in Children

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