



Empowering Children. Supporting Families. Building Brighter Futures.

Resource 070 | Genetic and chromosomal conditions

22q11.2 deletion syndrome

What it means

22q11.2 deletion syndrome is a genetic condition with a highly variable combination of medical and developmental features. It may affect the heart, palate, immune system, calcium regulation, communication, and learning. Some features emerge over time. Coordinated care is important even when only a few concerns are initially apparent. [1]

Causes and risk factors

The condition involves a missing segment of chromosome 22 containing several genes. Many cases occur as a new genetic change; some are inherited. Effects vary even within a family. Genetic counseling helps explain the result and family testing considerations without assuming a fixed developmental outcome. [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 congenital heart disease, feeding difficulties, nasal-sounding speech, palate differences, recurrent infections, low calcium, and developmental or learning difficulties. Hearing, attention, anxiety, and other mental health concerns may occur. A child does not need every characteristic to have the condition. [1,2]

Assessment and diagnosis

Genetic testing, often chromosomal microarray, identifies the deletion. Assessment covers cardiac, immune, endocrine, palate, feeding, hearing, and developmental needs. Clinical appearance alone is insufficient. Specialists determine the appropriate vaccination, infection, and procedural plans, which depend on the child's actual immune and medical findings. [1,2]

Treatment and therapy

Care addresses the organs and functions affected. Options include cardiology treatment, palate surgery when indicated, speech-language therapy, feeding support, and management of calcium or immune problems. Occupational and physical therapy, communication supports, and educational accommodations may help. A coordinated team also monitors emotional health and emerging learning needs over time. [1,2,3]

Support at home

Keep an up-to-date medical summary and follow the individualized infection and vaccination advice. Use accessible communication and support learning in small steps. Share feeding and speech plans across settings. Report changes in energy, attention, sleep, or participation, since these may reflect treatable health or emotional concerns.

When to seek help

Seek urgent help for severe infection, breathing difficulty, collapse, or seizures. New muscle spasms, marked lethargy, or poor feeding need prompt assessment, especially when low calcium is a known risk.

Questions for your care team

- Which organ systems need assessment and surveillance?
- What immune-specific vaccination and infection guidance applies?
- How should palate, speech, and learning supports be coordinated?



Podcast preparation

Use this outline to develop a later podcast script.

Suggested opening

Welcome to Mariel Early Intervention. Today we discuss 22q11.2 deletion syndrome. 22q11.2 deletion syndrome is a genetic condition with a highly variable combination of medical and developmental features. 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: every child has the same medical features. The deletion can produce very different needs.

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

Terms in plain language

- Chromosomal microarray: a test that detects certain missing or extra chromosome segments.
- Hypocalcemia: an abnormally low blood calcium level.
- Palate: the roof of the mouth, important for feeding and speech.

Sources and further reading

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

[1] 22q11.2 deletion syndrome: MedlinePlus Genetics

<https://medlineplus.gov/genetics/condition/22q112-deletion-syndrome/>

[2] 22q11.2 Deletion Syndrome - GeneReviews® - NCBI Bookshelf

<https://www.ncbi.nlm.nih.gov/books/NBK1523/>

[3] Assessing Developmental Delays in Children

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