



Empowering Children. Supporting Families. Building Brighter Futures.

#### **Resource 064 | Genetic and chromosomal conditions**

## **Down syndrome**

### **What it means**

Down syndrome is a genetic condition caused by extra chromosome 21 material. It affects development and is associated with particular medical risks, but each child's abilities and interests are individual. Early support combines health surveillance with communication, movement, self-care, and inclusive learning opportunities. [1]

### **Causes and risk factors**

Most cases involve three copies of chromosome 21 in the cells. Less commonly, extra chromosome material is attached to another chromosome or occurs in only some cells. Most cases are not inherited; some chromosome arrangements have family implications. Genetic counseling can explain the specific finding. [1,2]

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



## Signs and everyday impact

Low muscle tone and slower motor and language development are common. Heart conditions, hearing and vision differences, thyroid disease, feeding problems, and sleep-disordered breathing may affect participation. These concerns are not present in every child. Strengths and support needs should be assessed individually. [1,2]

## Assessment and diagnosis

Chromosome testing confirms the diagnosis. Prenatal screening estimates likelihood but is not itself a definitive diagnosis. After diagnosis, clinicians plan age-appropriate medical surveillance and developmental assessment. Hearing and vision access, communication, feeding, and sleep should be considered when interpreting learning or behavior. [1,2]

## Treatment and therapy

Treatment addresses associated medical conditions and individual developmental needs. Physical and occupational therapy can support movement and everyday skills; speech-language therapy and AAC support communication. Educational accommodations and inclusive opportunities help learning. Ongoing heart, hearing, vision, thyroid, and sleep care follow clinical guidance, with other specialists involved as needed. [1,2,3]

## Support at home

Use responsive conversation, shared books, gestures, and communication aids as appropriate. Give time for responses and break tasks into achievable steps. Support active play within medical advice. Keep health follow-up appointments because treatable hearing, sleep, or thyroid difficulties can affect learning and energy.

## When to seek help

Seek prompt assessment for new loss of skills, changed walking or neck-related symptoms, feeding deterioration, or suspected hearing loss. Breathing difficulty, blue coloring, or collapse requires emergency services.

## Questions for your care team

- Which health checks are due at this age?
- Would AAC improve communication now?
- How can therapy goals support participation and self-care?



## Podcast preparation

Use this outline to develop a later podcast script.

### Suggested opening

Welcome to Mariel Early Intervention. Today we discuss down syndrome. Down syndrome is a genetic condition caused by extra chromosome 21 material. 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: the diagnosis sets a fixed ceiling on learning. Individual assessment and accessible opportunities remain essential.

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

### Terms in plain language

- Trisomy: three copies of a chromosome instead of two.
- Mosaic: a genetic change present in some cells but not all.
- AAC: communication options such as signs, pictures, or devices.

### Sources and further reading

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

**[1] Down syndrome: MedlinePlus Genetics**

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

**[2] Down Syndrome**

<https://www.nichd.nih.gov/health/topics/downsyndrome>

**[3] What are common treatments for Down syndrome?**

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