



Empowering Children. Supporting Families. Building Brighter Futures.

Resource 028 | Movement and muscle conditions

Spinal muscular atrophy

What it means

Spinal muscular atrophy (SMA) is a group of genetic conditions affecting motor nerve cells and causing muscle weakness. The most common form is related to SMN1. Severity and age of onset vary. Early identification is important because available disease-modifying treatment is time-sensitive and requires specialist care. [1]

Causes and risk factors

Most childhood SMA results from changes in both copies of SMN1, reducing a protein needed by motor neurons. SMN2 and other factors influence the pattern, but do not predict every outcome. Other rarer forms of SMA have different genetic causes. Genetic counseling explains inheritance for the specific diagnosis. [1,2]

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



Signs and everyday impact

Infants may show reduced movement, low tone, poor head control, weak cry, feeding fatigue, or breathing difficulty. Older children may have trouble standing, walking, or rising from the floor. Weakness is often more prominent near the trunk. A child can have substantial movement difficulties without impaired understanding. [1,2]

Assessment and diagnosis

A pediatric neuromuscular team evaluates strength, reflexes, breathing, feeding, and development. Genetic testing usually confirms common SMN1-related SMA; other investigations depend on the findings. Newborn screening is available in some programs, but a previous screening result should not delay evaluation of concerning weakness. [1,2]

Treatment and therapy

Specialists may offer medicines or gene-based treatments that improve SMN-related function, according to diagnosis, age, eligibility, local approval, and medical risks. Respiratory, nutritional, orthopedic, and rehabilitation care remain important. Physical and occupational therapy, equipment, and communication support help participation. Prompt referral should not wait for routine therapy to produce progress. [1,2,3]

Support at home

Follow the respiratory, feeding, positioning, and illness plans. Allow rest and avoid overexertion. Ask about safe equipment and movement opportunities. Keep emergency information available and report changes in cough strength, intake, sleep, or movement promptly.

When to seek help

An infant with marked weakness, poor feeding, or reduced movement needs urgent assessment. Breathing difficulty, weak cough with illness, choking, or reduced responsiveness requires emergency care.

Questions for your care team

- Which genetic form has been confirmed?
- What time-sensitive treatment options and monitoring apply?
- What is our breathing, feeding, and illness plan?



Podcast preparation

Use this outline to develop a later podcast script.

Suggested opening

Welcome to Mariel Early Intervention. Today we discuss spinal muscular atrophy. Spinal muscular atrophy (SMA) is a group of genetic conditions affecting motor nerve cells and causing muscle weakness. 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: SMA treatment is not limited to exercises, and medical treatment does not remove the need for supportive care.

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

Terms in plain language

- Motor neuron: a nerve cell that signals muscles.
- SMN: a protein important to motor neuron survival.
- Disease-modifying: addressing a biological process of the condition.

Sources and further reading

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

[1] Spinal muscular atrophy: MedlinePlus Genetics

<https://medlineplus.gov/genetics/condition/spinal-muscular-atrophy/>

[2] Spinal muscular atrophy: MedlinePlus Medical Encyclopedia

<https://medlineplus.gov/ency/article/000996.htm>

[3] Hypotonia: MedlinePlus Medical Encyclopedia

<https://medlineplus.gov/ency/article/003298.htm>