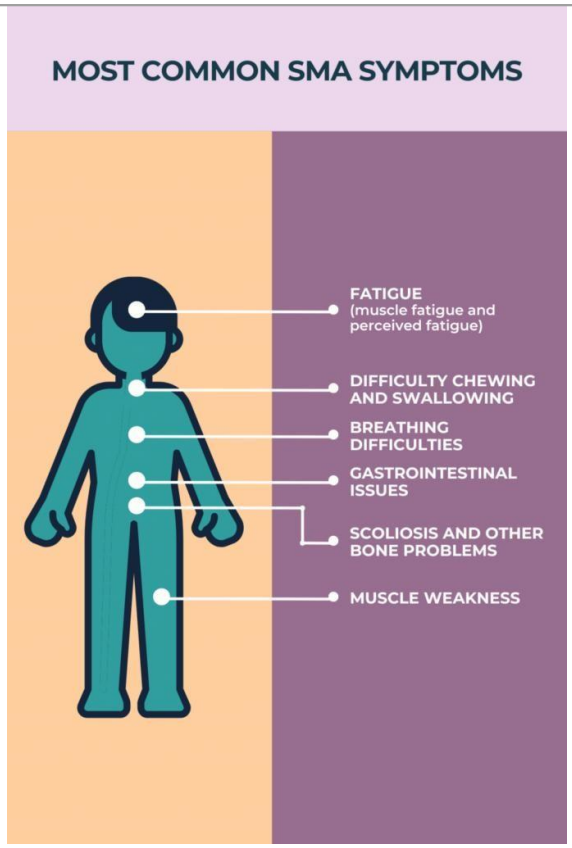
Spinal Muscular Atrophy (SMA)



What is Spinal Muscular Atrophy (SMA)?

Spinal Muscular Atrophy (SMA) is a **genetic neuromuscular condition** that affects the **motor nerves** in the spinal cord. These nerves control muscle movement. When they do not work properly, muscles become **weak and smaller (atrophy)** over time.

SMA mainly affects **movement muscles**, including those needed for crawling, walking, sitting, breathing, and sometimes swallowing.

What Causes SMA?

SMA is caused by a change (mutation) in a gene called **SMN1**.
This gene helps produce a protein needed for healthy motor nerve cells.

Children with SMA do not produce enough SMN protein, which leads to weakness and reduced motor function.

SMA is **inherited** in an **autosomal recessive** pattern—this means both parents must carry the gene change.

Types of SMA

SMA ranges from mild to severe. The main types include:

Type 1 (Severe)

- Symptoms appear in early infancy.
- Babies have weak muscles, difficulty holding head up, feeding, and breathing.

Type 2 (Intermediate)

- Symptoms start between 6–18 months.
- Children can sit but may not walk independently.

Type 3 (Mild)

- Symptoms start after 18 months or in childhood.
- Children can walk but may show weakness, frequent falls, or difficulty running and climbing.

Type 4 (Adult-Onset)

- Rare in children; begins in adulthood.

Every child with SMA is different—severity varies even within the same type.

Common Signs and Symptoms

Depending on the type, a child may have:

- Muscle weakness in legs, arms, or trunk
- Reduced muscle bulk
- Difficulty crawling, walking, or standing
- Delayed motor milestones

- Fatigue with activity
- Difficulty sitting or keeping good posture
- Scoliosis (curving of the spine) in some cases
- Breathing or feeding difficulties (more common in SMA types 1 & 2)

How is SMA Diagnosed?

Diagnosis usually includes:

- **Genetic testing** (to confirm SMN1 gene mutations)
- **Physical examination** • **Muscle strength assessment**
- **Electromyography (EMG)** or nerve conduction tests (in some cases) Early

diagnosis allows quicker access to treatment and support.

Treatment Options

There is currently **no cure** for SMA, but major advances mean most children benefit significantly from treatment.

Medical Treatments (via specialist teams):

- **Nusinersen (Spinraza)** – injected into the spine at intervals
- **Risdiplam (Evrysdi)** – taken orally
- **Onasemnogene abeparvovec (Zolgensma)** – a one-time gene therapy (for certain age/weight groups)

Your child's neuromuscular team will advise the best option based on type and individual needs.

Supportive Care

Management often involves a team including neurologists, physiotherapists, occupational therapists, speech therapists, and respiratory specialists.

Physiotherapy and Exercise

Physiotherapy is a key part of SMA management. It helps maintain:

- Joint flexibility
- Muscle strength
- Functional mobility

- Breathing efficiency
- Posture and comfort

Common physiotherapy strategies include:

- Gentle strengthening exercises
- Stretching to prevent tightness
- Postural support
- Aquatic therapy (hydrotherapy)
- Assistive devices (standing frames, walkers)
- Breathing exercises or chest physiotherapy

Daily Activities and How Parents Can Help

- Encourage regular **movement and play** within the child's ability.
- Monitor for signs of **fatigue**—rest breaks are important.
- Ensure safe seating and posture to protect the spine and breathing.
- Use mobility aids or braces if advised.
- Maintain good respiratory health (e.g., avoiding infections).

EXERCISES FOR SMA

Aqua therapy

Adaptive cycling

Yoga

Adaptive sports

Adaptive camps

Hippotherapy

PHYSIOTHERAPY

Aerobics

Resistance training

School and Social Support

Children with SMA can attend school and participate in most activities with appropriate support. This may include:

- Wheelchair access
- Extra time for physical tasks
- Support assistants
- Seating or posture aids
- Individual health plans

Your healthcare team can help with documentation and school coordination.

When to Seek Medical Advice

Contact your GP, physiotherapist, or neuromuscular team if your child has:

- Increasing weakness
- More difficulty breathing
- Feeding or swallowing problems
- Frequent chest infections
- Sudden loss of function

Seek urgent help if breathing becomes difficult.

Prognosis

With modern treatments and comprehensive care, many children with SMA can achieve good quality of life, improved mobility, and better long-term outcomes than previously expected.

Support Resources

- **SMA UK** – Information, support groups, and resources for families
- **Genetic counselling services** – For understanding inheritance and future planning
- **Local children's physiotherapy services** – Ongoing support and therapy programs