CONDITION

Spinal muscular atrophy

Spinal muscular atrophy is an inherited condition that weakens muscles used for movement, swallowing, and breathing.

WHAT IT IS
Inherited condition causing progressive muscle weakness and wasting

BODY SYSTEM
Nervous system and muscles

MAIN SYMPTOMS
Muscle weakness, low muscle tone, feeding difficulty, breathing problems

WHAT CAUSES IT
Inherited SMN1 gene changes reducing essential motor neuron protein

HOW IT IS DIAGNOSED
Genetic blood testing for SMN1 changes

MAIN TREATMENT
Disease-modifying therapy and multidisciplinary supportive care

CAN IT BE CURED?
No, treatments can greatly improve outcomes

IS IT AN EMERGENCY?
Sometimes, when breathing or swallowing suddenly becomes difficult

LONG-TERM OUTLOOK
Earlier treatment often improves movement, breathing, and survival

Overview

Spinal muscular atrophy, usually called SMA, is an inherited condition that causes muscle weakness and muscle wasting. It mainly affects motor neurons, the nerve cells in the spinal cord and lower brain that control voluntary movement.

When motor neurons do not work properly, muscles receive weaker signals. Over time, the muscles can become smaller and less able to move. SMA may affect sitting, standing, walking, swallowing, coughing, and breathing. It does not usually affect sensation or intelligence.

Most people with SMA have a form linked to the SMN1 gene. This is often called 5q SMA because the gene is located in a region of chromosome 5. A related gene, SMN2 , can make some of the protein that the body needs, but usually not enough to fully protect motor neurons.

SMA was traditionally divided into types 0 through 4 according to the age symptoms began and the strongest movement milestone reached. For example, type 1 begins in infancy, while type 4 begins in adulthood. These labels remain useful, but they are less able to predict the future now that disease-modifying treatments can change the condition's course.

Early diagnosis matters. Treatment started before symptoms, or soon after they appear, offers the best chance of protecting motor neurons that are still working.

Symptoms

SMA causes weakness that is usually similar on both sides of the body. Muscles close to the centre of the body, such as those in the shoulders, hips, and trunk, are often affected more than the hands and feet.

Possible symptoms include:

Symptoms and their timing differ widely. Some babies have weakness before birth or soon after delivery. Other children reach early movement milestones but later have difficulty standing or walking. People with milder forms may not notice symptoms until adolescence or adulthood.

SMA does not usually affect bladder or bowel control directly. Most people can feel touch, pain, and temperature normally. Thinking, learning, and emotional development are also usually unaffected.

- Low muscle tone, sometimes described as being "floppy"
- Difficulty lifting the head, rolling, sitting, standing, or walking
- Weak arms, legs, shoulders, or hips
- Reduced or absent reflexes
- Shaking or fine tremor in the fingers
- Muscle twitching in the tongue
- Difficulty chewing, swallowing, or gaining weight
- A weak cough or trouble clearing mucus
- Shortness of breath, especially during sleep or an infection
- Joint stiffness, curved spine, or changes in posture

Causes and risk factors

Most SMA is caused by changes in both copies of the SMN1 gene. This gene provides instructions for making survival motor neuron protein, which motor neurons need to remain healthy.

The usual inheritance pattern is autosomal recessive . This means a child inherits one altered SMN1 copy from each parent. Parents who have one altered copy are called carriers and usually do not have SMA themselves.

When both parents are carriers, each pregnancy has:

These chances start again with every pregnancy. They do not change based on the sex of the child or what happened in an earlier pregnancy.

The number of SMN2 gene copies can influence how much protein the body makes. More copies are often linked with milder symptoms, but the copy number cannot predict one person's course with certainty.

Rare forms of SMA are caused by changes in other genes and may follow different inheritance patterns. SMA is not caused by exercise, diet, infection, parenting, or anything someone did during pregnancy.

- A 1 in 4 chance of a child having SMA
- A 1 in 2 chance of a child being an unaffected carrier
- A 1 in 4 chance of a child inheriting neither altered copy

Diagnosis

In some places, SMA is included in newborn blood-spot screening. Screening can identify babies who may have SMA before weakness becomes visible. A positive screening result is not the final diagnosis and needs prompt confirmatory testing.

SMA is usually confirmed through a genetic blood test that looks for missing or altered copies of SMN1. Testing may also measure the number of SMN2 copies because this can help with treatment planning and discussions about likely severity.

If standard testing does not find the cause but SMA is still suspected, more detailed genetic testing may be needed. Tests such as electromyography, or EMG, can examine how nerves and muscles communicate. Blood tests may also help rule out other causes of weakness.

After diagnosis, assessments may cover movement, breathing, swallowing, nutrition, posture, bone health, and daily activities. Genetic counselling can help you understand inheritance,

testing for relatives, and reproductive options.

Treatment and management

SMA has no current cure, but disease-modifying treatment can protect motor neuron function and improve outcomes. Treatment is generally more effective when it begins early, before many motor neurons have been lost.

Available treatments include:

Eligibility and access differ by age, health, previous treatment, and local approval rules. Each treatment requires specific monitoring and can have important risks. Gene replacement does not remove the altered gene or restore motor neurons that have already been lost, so it is not considered a cure.

Supportive care remains important even when disease-modifying treatment is working. Care may involve specialists in neurology, breathing, rehabilitation, nutrition, speech and swallowing, orthopaedics, and genetics.

Breathing support can include assisted coughing, airway-clearance equipment, suction, or non-invasive ventilation through a mask. A respiratory plan is especially useful during colds and other infections.

A swallowing assessment can identify foods and drinks that are safer and easier to manage. Some people need nutritional supplements or feeding through a tube to maintain nutrition and reduce the risk of food entering the lungs.

Physiotherapy and occupational therapy can help preserve movement, comfort, flexibility, and independence. Stretching, braces, supportive seating, standing equipment, wheelchairs, and communication technology may also help. Exercise should be adapted to the person's strength and breathing ability rather than pushed to exhaustion.

- Nusinersen , which changes how the SMN2 gene is processed and is given into the fluid around the spinal cord
- Risdiplam , an oral medicine that helps SMN2 produce more working protein
- Onasemnogene abeparvovec , a one-time gene-replacement treatment given into a vein to eligible young children

Complications

Weak breathing muscles can make it difficult to take deep breaths, cough effectively, or clear

mucus. This can increase the chance of chest infections and breathing problems, particularly during sleep or acute illness.

Weakness in the mouth and throat can lead to choking or aspiration. Aspiration means food, drink, saliva, or stomach contents enter the airway. Repeated aspiration can contribute to lung infections.

Other possible complications include:

Regular monitoring is routine care, not a sign that a complication is certain. Identifying changes early can allow equipment, therapy, nutrition, or breathing support to be adjusted.

- Poor weight gain or difficulty meeting nutritional needs
- Excess weight when movement is limited
- Dehydration or constipation related to feeding difficulties
- Joint contractures, where a joint becomes fixed in a bent position
- Scoliosis, an abnormal sideways curve of the spine
- Hip instability, pain, or reduced mobility
- Reduced bone strength and fractures
- Fatigue and difficulty completing daily activities

Prevention

The inherited genetic changes that cause most SMA cannot be prevented through lifestyle choices.

Carrier screening can show whether someone carries an altered SMN1 copy, although testing has limitations and does not identify every carrier. If SMA runs in your family, genetic counselling can explain testing before or during pregnancy.

When the family's genetic changes are known, options may include prenatal diagnostic testing or in vitro fertilisation with testing of embryos. These are personal choices rather than ways to treat an existing pregnancy.

Newborn screening does not prevent SMA. It can identify the condition before symptoms begin, allowing treatment to start when it may provide greater benefit.

Living with it and what to expect

The course of SMA differs according to when symptoms begin, which muscles are affected, breathing and swallowing function, and how early effective treatment starts. Traditional SMA types describe the condition's untreated course more accurately than they predict outcomes after modern treatment.

Some people continue gaining movement skills after treatment. Others maintain existing abilities for longer or lose function more slowly. Treatment may not reverse established weakness, and ongoing support may still be needed.

Breathing, posture, swallowing, mobility, and nutrition can change over months or years. Regular follow-up helps track these areas and adapt support. Reviews may be more frequent in infancy, during rapid growth, after treatment changes, or when breathing and swallowing are changing.

Children with SMA can usually learn in mainstream education when suitable physical access and assistance are available. Adults may study, work, build relationships, parent, travel, and take part in their communities. Wheelchairs and other equipment can increase independence rather than represent a failure of treatment.

SMA can also affect family routines, sleep, finances, and emotional wellbeing. Practical support, mental health care, peer groups, respite services, and accessible education or employment arrangements may be useful. Because treatments are relatively new, their full long-term effects are still being studied.

When to get medical help

Arrange routine medical review if you notice new or gradually worsening weakness, increased fatigue, changes in walking or posture, weight loss, repeated coughing during meals, morning headaches, disturbed sleep, or more difficulty clearing mucus. These changes may mean that treatment, equipment, nutrition, or breathing support needs review.

Seek urgent medical help if you or your child has:

Call emergency services for severe trouble breathing, blue or grey lips or skin, a prolonged choking episode, loss of consciousness, or inability to speak or cry because of breathlessness.

A respiratory illness can become more serious when coughing muscles are weak. Follow any existing emergency or airway-clearance plan and tell emergency staff that the person has SMA.

- New difficulty swallowing saliva or repeated choking
- Breathing that is faster, shallower, or more difficult than usual

- A noticeably weaker cough during a respiratory infection
- Unusual sleepiness, confusion, or difficulty waking
- Reduced feeding or signs of dehydration during illness

Common questions

Is spinal muscular atrophy fatal?

Some severe forms can shorten life, mainly because of breathing complications. Disease-modifying treatments and improved respiratory and nutritional care have changed survival for many people. Individual outlook cannot be predicted from the SMA type alone.

Can someone with SMA learn to walk?

Some people walk independently, while others use braces, walkers, or wheelchairs. Whether walking develops or continues is influenced by the age symptoms began, strength before treatment, treatment timing, and the person's response.

Does SMA affect intelligence?

SMA does not usually affect intelligence. Physical weakness may make it harder to write, speak loudly, use classroom materials, or show what someone knows, so accessible equipment and enough time can be important.

Is SMA the same as ALS?

No. Both affect motor neurons, but they are different conditions. SMA is usually inherited and often begins in infancy or childhood, although adult-onset forms occur. Amyotrophic lateral sclerosis, or ALS, usually begins in adulthood and has different causes and patterns of nerve involvement.

Can carriers develop SMA?

People with one altered SMN1 copy are usually healthy carriers and do not develop classic 5q SMA. Genetic testing may be more complex when someone has symptoms or a family history, so specialist interpretation can be helpful.

Can SMA be detected before birth?

Yes, when the relevant family gene changes are known, diagnostic testing can be performed during pregnancy. Carrier screening estimates reproductive risk, while prenatal diagnostic testing determines whether the pregnancy has inherited the specific changes being tested.

Sources

- GeneReviews: Spinal Muscular Atrophy , Prior TW, Leach ME, Finanger E; University of Washington, Seattle
- Spinal muscular atrophy , NHS
- Mercuri E, Finkel RS, Muntoni F, et al. "Diagnosis and management of spinal muscular atrophy: Part 1." Neuromuscular Disorders . 2018;28(2):103-115
- Finkel RS, Mercuri E, Meyer OH, et al. "Diagnosis and management of spinal muscular atrophy: Part 2." Neuromuscular Disorders . 2018;28(3):197-207
- SPINRAZA prescribing information , U.S. Food and Drug Administration
- EVRYSDI prescribing information , U.S. Food and Drug Administration
- ZOLGENSMA prescribing information , U.S. Food and Drug Administration