CONDITION

Infantile spinal muscular atrophy type I

Infantile spinal muscular atrophy type I is an inherited condition that causes severe muscle weakness in babies by damaging nerve cells that control movement.

WHAT IT IS
Severe inherited loss of movement-controlling nerve cells

BODY SYSTEM
Nervous system and muscles

MAIN SYMPTOMS
Muscle weakness, poor head control, weak cry, breathing problems

WHAT CAUSES IT
Inherited changes in both SMN1 gene copies

HOW IT IS DIAGNOSED
SMN1 genetic testing, often after newborn screening

MAIN TREATMENT
Disease-modifying therapy plus breathing, feeding, and movement support

CAN IT BE CURED?
No, lost nerve cells cannot regrow

IS IT AN EMERGENCY?
Sometimes, with breathing difficulty, blue colour, or severe choking

LONG-TERM OUTLOOK
Often serious, but early treatment can greatly improve outcomes

Overview

Infantile spinal muscular atrophy type I is a severe inherited condition affecting babies. It damages motor neurons, the nerve cells in the brainstem and spinal cord that tell muscles to move.

As motor neurons are lost, muscles become weak and shrink. This can affect head control, movement, swallowing, coughing, and breathing. Thinking, awareness, sight, hearing, and sensation are not directly affected.

Symptoms usually begin during the first six months of life. In the traditional description of type I, babies do not achieve independent sitting without treatment. Newborn screening and early treatment are changing this pattern, so a baby's future abilities may not fit older descriptions of the condition.

This condition needs prompt specialist care because treatment works best before many motor neurons have been lost. Modern disease-modifying treatments can improve movement, reduce the need for breathing support, and extend survival. They cannot restore every nerve cell that has already been lost.

Symptoms

Some babies are identified through newborn screening before symptoms appear. Others are assessed after a parent or healthcare professional notices unusual weakness.

Possible signs include:

The weakness is usually similar on both sides of the body. The legs may be more affected than the arms. Facial expression and eye movement are generally preserved.

Breathing may look unusual because the muscles between the ribs are weak while the main breathing muscle under the lungs, called the diaphragm, continues working. The abdomen may move outward as the chest moves inward. This is called paradoxical breathing.

Small tremors of the fingers or twitching of the tongue can occur. These signs are not present in every baby and are not enough on their own to confirm the condition.

- Poor head control or difficulty lifting the head
- Reduced movement of the arms and legs
- A floppy feeling when the baby is held
- Difficulty feeding, sucking, or swallowing
- A weak cry or cough
- Fast, shallow, or laboured breathing
- A chest that appears bell-shaped
- Limited ability to meet movement milestones

Causes and risk factors

Infantile spinal muscular atrophy type I is usually caused by harmful changes in both copies of a gene called SMN1 . This gene provides instructions for making survival motor neuron, or SMN, protein. Motor neurons need this protein to remain healthy.

A nearby gene called SMN2 also makes some SMN protein, but most of it is incomplete. The

number of SMN2 copies can influence severity: more copies are often linked with milder symptoms. However, copy number does not predict an individual baby's course with certainty, especially after early treatment.

The condition follows an autosomal recessive inheritance pattern. This usually means both parents carry one altered SMN1 copy but do not have spinal muscular atrophy themselves. For each pregnancy between two carriers, the genetic outcome is determined separately.

Having no known family history does not rule it out. Carriers are usually healthy, and the altered gene can pass through a family unnoticed for generations.

Nothing a parent did during pregnancy causes infantile spinal muscular atrophy. It is not caused by an infection, an injury, or routine infant care.

Diagnosis

In places where spinal muscular atrophy is included in newborn screening, a heel-prick blood sample may show that the SMN1 gene is missing. Screening is not a final diagnosis, so an abnormal result needs rapid confirmatory testing.

Diagnosis is confirmed with genetic testing that looks for missing or harmful changes in both copies of SMN1. Testing may also measure the number of SMN2 copies to help the care team discuss likely severity and treatment planning.

If symptoms appear before screening results are available, genetic testing should not be delayed. A specialist will also assess muscle tone, movement, reflexes, breathing, feeding, and developmental milestones.

Most babies do not need a muscle biopsy. Tests of nerves and muscles, such as electromyography, may be considered when the genetic result is unclear or another condition is possible.

Parents and sometimes siblings may be offered genetic testing after a diagnosis. A genetic counsellor can explain what the result means for other family members and future pregnancies.

Treatment and management

Treatment is coordinated by a specialist neuromuscular team. Starting disease-modifying therapy as early as possible is important because treatment can protect remaining motor neurons but cannot replace all those already lost.

Available treatments work in different ways:

Which treatments are available or suitable is influenced by local approval, age, weight, previous treatment, test results, and the baby's overall health. Each option requires specific monitoring. The specialist team will discuss expected benefits, uncertainties, and treatment-related risks.

Supportive care remains important even when disease-modifying treatment is working. Breathing care may include help clearing mucus, assisted coughing, suction, or non-invasive ventilation through a mask. The team may monitor breathing during sleep because weakness can first become noticeable then.

A feeding and swallowing assessment can identify whether milk or food is entering the airway. Changes to feeding position or texture may help. Some babies need temporary or longer-term tube feeding to provide nutrition safely.

Physiotherapy and occupational therapy support comfortable positioning, joint movement, play, and access to equipment. Gentle movement may help prevent stiffness, but therapy should avoid exhausting weak muscles.

Palliative care may be offered alongside active treatment. It focuses on comfort, symptom relief, family support, and decisions that reflect the family's priorities. It does not mean that other treatments must stop.

- Nusinersen changes how the SMN2 gene is used so that it makes more working SMN protein. It is given into the fluid around the spinal cord at scheduled intervals.
- Onasemnogene abeparvovec is a one-time gene-replacement treatment that delivers a working copy of the SMN1 gene into cells.
- Risdiplam is taken by mouth and helps SMN2 produce more working SMN protein.

Complications

Weak breathing muscles make it difficult to take deep breaths and clear mucus. A cold or other respiratory infection can therefore cause more difficulty than it would in another baby. Some children need ongoing ventilation support, especially during sleep.

Swallowing weakness can cause choking or aspiration, which means milk, food, or saliva enters the airway. Aspiration can contribute to chest infections and may occur without obvious coughing.

Poor feeding can lead to slow weight gain, while reduced movement can affect bone strength. Careful nutrition planning is needed because both undernutrition and excess weight can make breathing and movement harder.

Over time, muscle weakness can contribute to tight joints, hip problems, or curvature of the spine. Seating, positioning, movement therapy, and orthopaedic monitoring can help maintain comfort and function.

Treatment has improved survival, so clinicians are learning more about the long-term needs of children who would previously not have lived as long. Follow-up may include movement, breathing, swallowing, nutrition, growth, spine health, and development.

Prevention

The genetic changes that cause infantile spinal muscular atrophy type I cannot be prevented in a baby who has inherited them. Parents are not responsible for causing the condition.

Carrier testing can show whether someone has an increased chance of having a child with spinal muscular atrophy. When both prospective parents are carriers, a genetic counsellor can explain reproductive options. These may include testing during pregnancy or testing embryos created through in vitro fertilisation.

These choices are personal. Testing provides information, but it does not remove every uncertainty or require a particular decision.

Newborn screening does not prevent spinal muscular atrophy. Its purpose is to identify affected babies quickly so treatment can begin before symptoms develop or progress.

Living with it and what to expect

The course of infantile spinal muscular atrophy type I has changed substantially with newborn screening and disease-modifying treatment. Babies treated before symptoms appear often achieve more movement milestones than older descriptions of type I would suggest. Treatment after symptoms begin may still improve movement, breathing, and survival, but response is less predictable.

Some children learn to sit, stand, or walk with or without support. Others continue to have major weakness and need powered mobility, breathing assistance, or tube feeding. Gains may occur slowly over months. Skills already lost can be harder to recover.

Treatment does not remove the underlying need for monitoring. Children usually have regular appointments with neuromuscular, respiratory, nutrition, rehabilitation, and therapy teams. Some therapies continue long term, while the effects of one-time gene replacement still require follow-up.

Illnesses may temporarily reduce strength or breathing ability. Families often receive a written plan for respiratory infections, equipment use, and emergency care. Vaccination and routine child healthcare remain important and should be discussed with the child's team.

Children with this condition can communicate, learn, build relationships, attend school, and take part in family and community life. Adapted seating, communication tools, mobility equipment, and personal assistance can support independence. The level of support needed differs between children and may change as they grow.

Long-term outcomes with newer treatments are still being studied. The specialist team can describe what has changed since the previous visit without promising a particular developmental path.

When to get medical help

Call emergency services now if your baby:

Seek urgent same-day medical advice for increased work of breathing, a much weaker cough, repeated choking, fever with breathing changes, unusual sleepiness, or signs of dehydration such as fewer wet nappies. Babies with spinal muscular atrophy can worsen quickly during respiratory infections.

Contact the specialist team promptly if feeding takes much longer, weight gain slows, coughing occurs during feeds, sleep becomes disturbed by breathing, or movement skills decrease.

Routine follow-up is used to review treatment response, growth, swallowing, respiratory support, equipment, and movement. New concerns do not need to wait until the next scheduled appointment.

- Is struggling severely to breathe or stops breathing
- Has blue, grey, or unusually pale lips or skin
- Is choking and cannot breathe, cry, or cough
- Is very difficult to wake or becomes unresponsive
- Has a sudden major decline in breathing or alertness

Common questions

Is infantile spinal muscular atrophy type I curable?

There is currently no cure that restores all lost motor neurons or removes the inherited gene change from every cell. Disease-modifying treatment can raise SMN protein levels or provide a working SMN1 gene, which may greatly improve survival and function.

Will my baby be able to sit or walk?

It is not possible to predict this from the diagnosis alone. Some babies treated very early achieve independent sitting or walking. Others need support for these activities. Timing of treatment, weakness already present, SMN2 copy number, and individual response all play a part.

Does spinal muscular atrophy affect intelligence?

It does not directly damage the parts of the brain responsible for thinking and understanding. Severe weakness can limit how a child moves, speaks, or shows what they know, so suitable communication and access tools are important.

Can my baby receive more than one SMA treatment?

Some children receive treatment after previously having another therapy, but evidence about treatment combinations and sequences is still developing. Decisions require specialist review of potential benefit, uncertainty, monitoring needs, and treatment-related risks.

Will another child in the family have it?

When both parents carry an altered SMN1 copy, each pregnancy has its own chance of being affected. Genetic testing can clarify carrier status and inheritance within the family. A genetic counsellor can explain the result without directing reproductive decisions.

Why does early treatment matter so much?

Motor neurons lost to the condition cannot be fully replaced. Treating before symptoms, or as soon as possible after they begin, gives therapy the best opportunity to preserve nerve cells and muscle function.

Sources

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