CONDITION

Infantile spinal muscular atrophy

Infantile spinal muscular atrophy is an inherited condition that causes severe muscle weakness beginning early in life.

WHAT IT IS
Inherited loss of motor neurons causing infant muscle weakness

BODY SYSTEM
Nervous system and muscles

MAIN SYMPTOMS
Floppiness, weak movement, feeding trouble, breathing weakness

WHAT CAUSES IT
Inherited SMN1 gene changes

HOW IT IS DIAGNOSED
Genetic testing confirming missing or altered SMN1 genes

MAIN TREATMENT
SMN-targeted therapy plus breathing, feeding, and movement support

CAN IT BE CURED?
No - treatment cannot restore all lost motor neurons

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

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

Overview

Infantile spinal muscular atrophy, or infantile SMA, is an inherited condition affecting the nerves that control voluntary muscles. It is usually caused by changes in a gene called SMN1 .

The affected nerves are called motor neurons. They carry instructions from the spinal cord to muscles used for movement, swallowing, coughing, and breathing. When these neurons do not receive enough survival motor neuron, or SMN, protein, they become damaged and are lost. The muscles then become weak and shrink from lack of use.

Infantile SMA commonly refers to SMA that begins before 6 months of age. It has traditionally been called SMA type 1. Without treatment, babies with this form usually do not develop the ability to sit independently. Modern treatments can change this pattern, especially when started before symptoms or soon after they appear. For this reason, older SMA "types" do not

always predict what a treated child will be able to do.[1]

SMA affects muscle strength but does not usually affect sensation. Babies are generally alert and interested in people and their surroundings. Intelligence is not thought to be directly affected.

Early diagnosis matters because treatment cannot replace motor neurons that have already been lost. Newborn screening now identifies some babies before weakness becomes obvious.

Symptoms

Symptoms may be present at birth or develop during the first months of life. They can become clearer as a baby misses movement milestones or loses strength.

Possible signs include:

The legs are often weaker than the arms. The muscles controlling eye movement are usually unaffected, and facial expression may remain lively.

Feeding may take a long time. Milk may leak from the mouth, or a baby may cough, choke, become tired, or breathe differently during feeds. Some babies silently breathe milk or saliva into their lungs without obvious coughing. This is called aspiration.

Breathing weakness may first become noticeable during sleep or a respiratory infection. A weak cough makes it harder to clear mucus, which raises the risk of chest infections and partially collapsed areas of lung.

These symptoms can have other causes. Genetic testing is needed to confirm SMA.

- Low muscle tone, sometimes described as being unusually floppy
- Less movement of the arms and legs
- Difficulty holding up the head
- Weak crying, coughing, or sucking
- Trouble swallowing or taking enough milk
- Slow weight gain
- Fast, shallow, or laboured breathing
- A chest that looks bell-shaped
- Breathing in which the abdomen rises while the chest moves inward
- Small trembling movements of the tongue

- Reduced reflexes

Causes and risk factors

Most infantile SMA is caused by missing or non-working copies of the SMN1 gene. This is known as 5q SMA because SMN1 is located in a region of chromosome 5 called 5q.

SMA is usually inherited in an autosomal recessive pattern. This means a child has received a non-working SMN1 copy from each parent. The parents are usually healthy carriers and may not know they carry the gene change.

When both parents are carriers, each pregnancy has:

These chances start again with every pregnancy. Occasionally, a new genetic change or a more complex gene pattern is involved.

A second gene, SMN2 , produces a smaller amount of working SMN protein. Having more SMN2 copies is often linked with milder symptoms, but the copy number cannot predict an individual child's course with certainty.

The main risk factor is having two carrier parents. A known family history increases the chance that carrier testing will identify risk, but SMA can occur without any known family history. It is not caused by anything a parent 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 having neither altered copy

Diagnosis

In places with newborn screening for SMA, a small blood sample taken soon after birth may show that the usual working part of SMN1 is missing. Screening is not the final diagnosis. A positive result needs prompt confirmatory genetic testing.

Diagnostic testing usually measures how many copies of SMN1 are present. If this does not explain strong clinical signs, sequencing may look for less common SMN1 changes. Testing also usually measures the number of SMN2 copies because this can help the care team discuss likely severity and treatment urgency.[1]

A physical examination assesses muscle tone, strength, reflexes, breathing, feeding, and movement milestones. Blood tests or electrical tests of muscles and nerves may be used

when the genetic result is unclear or another condition is possible. They are not usually needed to confirm a straightforward SMN1-related diagnosis.

Parents may be offered carrier testing. A genetics professional can explain the result, inheritance pattern, and what it may mean for siblings and future pregnancies.

Treatment and management

Treatment should begin as early as possible after a confirmed diagnosis. Babies identified before symptoms often have better motor outcomes than those treated after substantial weakness has developed. Treatment response is not identical for every child.

Three types of treatment can increase the amount of working SMN protein:

Which options are available depends on the country, the child's age and health, previous treatment, and current medicine approvals. These treatments have different risks and monitoring needs. Monitoring may include liver, kidney, blood-clotting, or platelet tests, depending on the treatment.[4][5][6]

Treatment that targets SMN does not remove the need for supportive care. Infantile SMA is best managed through a team experienced in neuromuscular conditions.

Breathing support may include assisted coughing, mucus clearance, suction, and non-invasive ventilation through a mask. Families often receive a plan for respiratory infections. Oxygen may sometimes be needed, but it does not by itself correct weak breathing muscles.

Feeding care may involve a swallowing assessment, changes in feeding position or texture, and nutrition support. If swallowing is unsafe or feeding takes too much energy, milk and medicines may be given through a tube into the stomach.

Physical and occupational therapy help protect comfortable movement and joint flexibility. Supportive seating, splints, standing equipment, and mobility devices may help a child participate in daily life. Treatment goals include comfort, communication, play, development, and family priorities, not only motor milestones.[2][3]

Palliative care can also be involved at any stage. It supports comfort, symptom control, and family decision-making and can be provided alongside treatments intended to change the condition's course.

- Nusinersen changes how the SMN2 gene is used. It is given into the fluid around the spinal cord through a lumbar puncture.
- Risdiplam also changes SMN2 activity. It is taken by mouth.

- Onasemnogene abeparvovec supplies a working copy of the SMN1 gene through a one-time intravenous infusion.

Complications

Weak breathing muscles can lead to poor ventilation, especially during sleep. Ventilation means moving enough air into and out of the lungs. Respiratory infections may become more serious because a weak cough cannot clear mucus effectively.

Swallowing weakness can cause dehydration, poor growth, or aspiration. Repeated aspiration can irritate the lungs and contribute to infection.

Limited movement may cause joints to become fixed in one position. These changes are called contractures. Curvature of the spine, known as scoliosis, may develop as a child grows. Fragile bones and fractures can also occur when muscles and bones do not bear much weight.

Long periods of illness and intensive care can affect sleep, family routines, finances, and emotional health. Social work, psychology, respite care, and parent support organisations can be useful parts of care.

Prevention

Infantile SMA cannot be prevented in a child who has inherited two non-working SMN1 copies. Exercise, diet, vitamins, and pregnancy choices do not prevent the genetic change.

Carrier screening can identify some people at increased chance of having a child with SMA. If both prospective parents are carriers, reproductive options may include prenatal testing or testing embryos created through in vitro fertilisation. These are personal choices that can be discussed with a genetics professional.

Newborn screening does not prevent SMA. It can allow treatment before symptoms appear, which may preserve more motor function.

Living with it and what to expect

Infantile SMA has historically been a life-limiting condition, mainly because of breathing weakness. SMN-targeted treatments and improved respiratory and nutrition care have changed the outlook. More children now survive longer and achieve abilities that were uncommon in untreated infantile SMA.[1]

The course over months and years remains difficult to predict for one child. It is influenced by how early treatment began, how many motor neurons were preserved, SMN2 copy number, respiratory health, feeding safety, and response to treatment.

Some treated children learn to sit, stand, or walk. Others continue to need extensive support for movement, breathing, and nutrition. Treatment may stabilise or improve certain abilities, but existing weakness may remain. Long-term information about children treated before symptoms is still developing.

Regular follow-up usually includes checks of breathing, swallowing, growth, movement, spine shape, joint flexibility, and treatment safety. These planned appointments are important even when a child seems stable because breathing or swallowing problems may not be obvious at first.

Children with SMA can communicate, learn, form relationships, attend school, and take part in family and community life. Adaptive equipment and accessible technology can support independence. Care plans may change as the child grows, but the genetic condition itself does not spread to other parts of the body or become contagious.

When to get medical help

Call emergency services now if your baby or child:

Seek urgent same-day medical advice for a weaker cough, increasing mucus, repeated choking, much less feeding, fewer wet diapers, fever with breathing changes, or greater effort needed to breathe. Respiratory infections can worsen muscle weakness quickly.

Contact the child's neuromuscular team promptly about a new decline in movement, feeding that regularly causes coughing or exhaustion, poor weight gain, morning headaches, disturbed sleep, or increasing sleepiness. These changes need assessment but are not always emergencies.

Routine follow-up should continue as planned for breathing tests, nutrition review, therapy, equipment, growth, and medicine monitoring.

- Is struggling to breathe, gasping, or having pauses in breathing
- Has blue, grey, or unusually pale lips or skin
- Is choking and cannot cough, cry, or breathe
- Is very difficult to wake or becomes unresponsive
- Has a sudden major loss of strength with breathing difficulty

Common questions

Is infantile spinal muscular atrophy fatal?

It can be life-limiting, especially when severe breathing weakness is untreated. Current treatments and respiratory support have improved survival, but outcomes differ between children. The timing of diagnosis and treatment is especially important.

Can a baby with SMA learn to sit or walk?

Some babies treated early learn to sit, stand, or walk. Others do not reach these milestones or need support to do so. A genetic result or SMN2 copy number cannot guarantee a particular outcome.

Is infantile SMA a form of muscular dystrophy?

No. Both conditions cause muscle weakness, but SMA begins with loss of motor neurons in the spinal cord. Muscular dystrophies mainly begin with damage within the muscle itself.

Does SMA affect intelligence?

SMA does not usually directly affect intelligence. Weakness can limit movement, speech volume, or access to activities, so a child may need adapted ways to communicate and participate.

Can gene therapy cure SMA?

Gene replacement therapy can provide a working SMN1 gene and may greatly change the condition's course. It is not considered a cure because it cannot restore motor neurons already lost, and children may continue to need monitoring and supportive care.

Could another child in the family have SMA?

If both parents carry a non-working SMN1 copy, each pregnancy usually has a 1 in 4 chance of SMA. Carrier and genetic testing can clarify the family's specific situation.

Sources

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