

# Spinal Muscular Atrophy and the Spine

A patient guide

**In brief:** Spinal muscular atrophy, or SMA, is an inherited disorder of the motor nerve cells in the spinal cord, caused in almost all cases by a fault in a gene called SMN1. The loss of these motor neurones leaves the muscles weak and, over time, wasted. While SMA is fundamentally a neuromuscular condition, it has a profound and very particular effect on the spine, and it is that spinal dimension that this page addresses. The weakness of the muscles that hold the trunk upright means that the growing spine has little to support it, and a curvature, a scoliosis, develops in the great majority of children with the more severe forms. This is a neuromuscular scoliosis, and it behaves quite differently from the idiopathic curves seen in otherwise healthy adolescents: it tends to appear early, to progress relentlessly, to form a long sweeping C-shaped curve, and to drag the pelvis out of level. The advent of disease-modifying drugs such as nusinersen, given by injection into the fluid around the spinal cord, has changed the natural history of the disease and has added a new and important consideration to any decision about spinal surgery, because a spine that is fused must still allow that medicine to be delivered. The single most important idea is that in SMA the spine is shaped by the weakness of the muscles around it, and that managing the spine is about supporting a trunk the muscles can no longer hold.

## What spinal muscular atrophy is, seen from the spine

Spinal muscular atrophy is a genetic condition in which the specialised nerve cells that drive the muscles, the lower motor neurones in the front of the spinal cord, are progressively lost. In almost all cases this stems from the loss of a working copy of a gene called SMN1, which produces a protein essential for the survival of these neurones. A second, nearly identical gene, SMN2, produces a small amount of the same protein, and the number of copies of SMN2 a person carries strongly influences how severe the disease is. When the motor neurones fail, the muscles they supply are no longer activated, and they become weak and wasted.

The condition is traditionally divided into types according to the age at which it begins and the milestones a child achieves. Type I, the most severe, begins in the first few months of life in infants who never sit unaided. Type II begins in later infancy in children who can sit but never walk independently. Type III begins after the child has learnt to walk, and type IV is a rare adult-onset form. This gradation matters enormously for the spine, because the earlier and more severe the weakness, the earlier and more aggressive the scoliosis tends to be.

Seen from the spine, the essential point is mechanical. The vertebral column is a flexible stack of bones that, in a healthy person, is held erect and balanced by a cradle of trunk muscles working constantly and almost unconsciously. In SMA those muscles are weak. The spine is left, in effect, without its scaffolding, and a young, growing, flexible spine that is not held straight will not stay straight. It is this loss of muscular support, rather than any primary fault in the bones or discs themselves, that drives the spinal deformity of SMA.

## Why scoliosis is so common in spinal muscular atrophy

Scoliosis is the dominant spinal problem in SMA, and in the more severe types it is very nearly universal. In children with type II disease, who can sit but not stand, and in many with the weaker end of type III, a progressive curvature of the spine develops in the great majority, and the published natural-history literature describes scoliosis in most non-ambulant children, often appearing within the first years of life. The weaker the child and the earlier the loss of the ability to stand and walk, the earlier and more severe the curve.

The reason follows directly from the mechanics already described. A child who cannot stand spends most of the day seated, and a trunk that cannot be held erect by its own muscles tends to collapse to one side under gravity.

Without the constant balancing work of the paraspinal and trunk muscles, small asymmetries are not corrected, and the spine bends progressively further. Because the deformity is driven by weakness acting on a growing spine throughout childhood, it characteristically begins early and continues to worsen so long as the child is growing, often accelerating sharply during the adolescent growth spurt.

This is what distinguishes a neuromuscular scoliosis from the more familiar adolescent idiopathic scoliosis. Idiopathic curves arise in otherwise healthy and well-muscled adolescents, tend to involve specific regions of the spine, and many never reach a magnitude that requires treatment. Neuromuscular curves of the kind seen in SMA arise from a global failure of trunk support, begin earlier, progress more reliably and to greater magnitudes, and very often involve the pelvis as well as the spine. They are, in essence, a structural consequence of profound muscle weakness.

## The shape of the curve: collapsing C-shaped deformity and pelvic obliquity

The scoliosis of SMA has a characteristic shape, and understanding it explains much of why it is treated as it is. Where idiopathic curves are often shorter and confined to the thoracic or lumbar region, the neuromuscular curve in SMA is typically a long, sweeping, single C-shaped curve that runs through the thoracic and lumbar spine together. It is sometimes described as a collapsing curve, because the trunk has, in effect, folded to one side for want of muscular support.

A defining feature of this pattern is that the curve does not stop at the spine but extends down into the pelvis. The base of the spine is no longer level, and one side of the pelvis is carried higher than the other. This is pelvic obliquity, and it is one of the most important consequences of the deformity. When the pelvis is tilted, the child sits unevenly, with the weight borne disproportionately on one side. This uneven sitting is uncomfortable, makes balanced seating in a wheelchair difficult, and concentrates pressure over the lower-lying side, where the skin is at risk. Restoring a level pelvis and a balanced sitting position is therefore one of the central aims when the deformity is addressed surgically.

Alongside the sideways curve there is often a loss of the normal front-to-back curves of the spine. The deformity is three-dimensional: the vertebrae rotate as they curve, and the rib cage on the concave side is drawn together while the ribs on the convex side splay apart. As the curve worsens, the trunk shortens and the lower ribs may come to rest close to, or against, the rim of the pelvis. The result is a foreshortened, collapsed and tilted trunk, and it is this whole picture, rather than the angle on an X-ray alone, that determines how the deformity affects a child's comfort, seating and breathing.

## How the spine and the lungs are connected in SMA

The spine and the lungs cannot be considered separately in SMA, and the respiratory dimension runs through every aspect of the spinal problem. The same weakness that fails to support the spine also weakens the muscles of breathing, particularly the intercostal muscles between the ribs, while the diaphragm is relatively spared. Children with the more severe forms therefore have a degree of respiratory weakness from the outset, quite apart from anything the spine does.

The deformity then makes this worse. As the thoracic spine curves and the vertebrae rotate, the rib cage is distorted: it is compressed on the concave side and the chest loses the room in which the lungs can expand. A severe, collapsing thoracic curve reduces the volume available to the lungs and the efficiency with which the chest wall can move air. In a growing child this is compounded by the fact that the thorax is still developing, and a spine that is severely deformed during the years of growth can impair the growth of the lungs themselves, a problem sometimes described in terms of thoracic insufficiency. The published surgical literature consistently records that restrictive lung disease is common in these children and that pulmonary function tends to decline over time.

This connection between the spine and the lungs shapes the whole approach to the deformity. Respiratory weakness is a major part of the risk of any operation on the spine in SMA, and the state of a child's breathing

weighs heavily in any decision about surgery, its timing and its conduct. At the same time, the hope that controlling the deformity may help preserve the space available for the lungs is one of the reasons the deformity is addressed at all. The spine and the chest are, in this condition, two aspects of a single problem.

## Disease-modifying therapy and why it changes the spinal picture

For most of the history of SMA there was no treatment that altered the underlying disease, and the spinal deformity ran its natural course. That has changed with the arrival of disease-modifying therapies, and this has had two distinct effects on the spinal picture that are important to hold in mind together.

The first effect is on the children themselves. Drugs that increase the amount of the missing SMN protein, including nusinersen, have improved survival and motor function, particularly in those treated early. More children with the severe forms now survive into and through childhood, and some achieve more than the natural history would have predicted. A direct consequence is that more children live long enough to develop, and to need attention for, a scoliosis that in earlier eras they might not have survived to develop. The spinal deformity has, in this sense, become a more prominent part of the condition precisely because the children are doing better.

The second effect is more particular and is one of the most distinctive considerations in SMA spine surgery. Nusinersen is not a tablet; it is given by injection into the cerebrospinal fluid in the lower back, by a lumbar puncture, repeated at intervals over years. A normal lumbar puncture passes a needle between the bony arches of the vertebrae into the spinal canal. A scoliosis distorts that anatomy and makes the puncture more difficult, and a spinal fusion, which locks the vertebrae together with bone and metal, can close off the very route the needle must take. The need to keep delivering this medicine through a spine that may need to be fused has become a genuine and recurring consideration in planning surgery, and the literature describes a number of techniques developed specifically to preserve that access.

## Preserving access for intrathecal medicine through a fused spine

The problem of delivering nusinersen into a spine that has been fused has prompted a body of practical work, and it is worth describing because it is so specific to the modern era of SMA. When the lumbar spine is fused with bone graft and rods, the spaces between the vertebral arches through which a lumbar puncture would normally pass are bridged over, and a needle inserted blindly between the rods may have no clear path to the fluid. Several approaches have been developed to address this.

One approach is surgical. At the time of the fusion, or as a separate procedure, a small window can be left or created in the bone of the lower lumbar spine, below the end of the spinal cord and between the rods, through which a needle can later be passed into the fluid. This has been described variously as a laminotomy or laminectomy window, or as a unilateral interlaminar fenestration, and the published technical accounts report that injections can be delivered through such a window over many years while the mechanical stability of the fusion is preserved. The window is placed low, below the level at which the spinal cord ends, to keep the puncture away from the cord itself.

A second approach is to rely on image guidance to find a path that already exists. Real-time ultrasound can be used to identify a gap between the bony layers and to steer the needle into it without radiation, an approach particularly suited to children. Where the bony anatomy is more complex, computed tomography or a cone-beam scan in the procedure room can be used to plan and guide the needle, sometimes by an unconventional route through the side of the spine, exploiting the way the bones are displaced by the scoliosis. The published series describe high rates of successful delivery by these means. The essential point is that, in the era of disease-modifying therapy, the design of a spinal fusion in SMA increasingly takes into account not only how to correct the deformity but how the spine will be accessed for years afterwards.

## Growth-friendly options and definitive fusion

When the deformity in SMA comes to be treated surgically, a central tension runs through the decision: the child is young and still growing, yet the spine needs support that, in its most durable form, means locking the vertebrae together. Fusing a young spine stops it growing in length over the fused segment, which can limit the growth of the trunk and the chest at an age when both matter for the lungs. Two broad strategies have developed in response to this, and the choice between them turns largely on the age and size of the child.

In younger children, growth-friendly techniques aim to control the curve while allowing the spine and chest to keep growing. The classic form is the growing rod, in which rods are anchored to the spine, or to the spine and the pelvis, without fusing the vertebrae in between, and are lengthened periodically as the child grows. Older systems required a small operation for each lengthening; magnetically controlled growing rods can be lengthened non-invasively, with an external device, sparing the child repeated surgery. The published experience in SMA reports that growing rods can reduce the curve and the pelvic obliquity, improve the height of the trunk and the space available for the lungs, and allow continued growth, although they do not arrest every aspect of the deformity, such as the collapse of the rib cage, and they carry their own burden of complications over the years of repeated lengthening.

In older children, in whom most growth is complete, a definitive posterior spinal fusion is the more durable solution. The spine is straightened as far as is safe, instrumented with screws, hooks and rods, and fused with bone graft so that it becomes a single solid column held permanently in the corrected position. In the non-ambulant child with a curve that reaches into the pelvis, the fusion is usually carried down to the pelvis itself, fixing the level of the sitting base. The surgical series report that fusion reliably corrects the curve and the pelvic obliquity and provides lasting, balanced support, and that it improves sitting comfort and seating, while acknowledging that it is major surgery in a fragile child. The transition from a growth-friendly construct in early childhood to a definitive fusion at maturity is a recognised pathway, and the relative merits of starting with one or the other are an active subject in the literature.

## Major surgical considerations in the SMA spine

Surgery on the spine in SMA is among the more demanding undertakings in deformity surgery, not because the correction itself is unusual but because of the fragility of the child and the particular features of the disease, and several considerations recur throughout the published accounts.

**\*\*Respiratory weakness dominates the risk.\*\*** The same muscle weakness that causes the deformity weakens breathing, and many children have restrictive lung disease before any operation. Respiratory complications are among the most frequent after major spine surgery in SMA, and careful assessment and support of breathing before, during and after the operation is a constant theme. The state of a child's lungs is one of the principal factors weighed in deciding whether, when and how to operate.

**\*\*The bone is often soft.\*\*** Disuse and immobility leave the bones thin, and weak bone holds screws and hooks less securely, so the fixation of metalwork into the spine and pelvis must contend with poor bone quality. This influences the choice and number of anchor points and the design of the construct.

**\*\*Blood loss, nutrition and positioning all require attention.\*\*** These long operations on the whole length of the spine can involve significant blood loss, and children with SMA are often slight, with limited reserves and frequently with poor nutrition and low body weight, all of which affect how well they tolerate and recover from surgery. The contracted joints and fragile skin of a child who has spent years seated also demand careful handling and positioning.

**\*\*Access for future medicine is now part of the plan.\*\*** As described above, the need to deliver intrathecal therapy for years afterwards has become a genuine part of surgical planning, influencing how low the fusion extends and whether a window for future injections is preserved.

**\*\*The aims are framed around the whole child.\*\*** The purpose of surgery in this setting is not cosmetic and is rarely framed simply in terms of the angle of the curve. It is to provide a balanced, level, comfortable sitting position, to relieve pressure on the skin, to free the hands and arms from the work of propping up the trunk, to support the chest, and to do so as safely as the child's fragility allows. These goals, and the risks set against them, are the substance of the careful, individualised discussions that surround any such operation.

## Outlook

The outlook in the spine of a child with SMA has changed substantially in recent years, and it can only be understood against the backdrop of the disease as a whole. For the severe forms, the arrival of disease-modifying therapy has improved survival and function, and as a result more children grow up to face the spinal deformity that profound weakness produces. The scoliosis itself remains, in the more severe types, a very common and progressive problem, and for most affected children the question is not whether a curve will develop but how it will be supported as they grow.

Where the deformity is treated, the surgical literature is, on the whole, encouraging about what can be achieved. Growth-friendly rods in younger children can control the curve and the pelvic obliquity while allowing growth to continue, and definitive fusion at maturity reliably restores a balanced, level and durable sitting position. These operations are recorded as improving sitting comfort, seating and the support of the trunk, even as the published series are equally honest about the high rate of complications, the burden of repeated procedures with growing systems, and the respiratory fragility that shadows the whole enterprise.

Running through the whole subject are a few consistent themes: that the deformity is a structural consequence of muscle weakness acting on a growing spine, that the spine and the lungs are inseparable in this condition, that disease-modifying medicine has both increased the relevance of the spinal problem and added the new demand of preserving access through a fused spine, and that every decision is finely balanced and deeply individual. The shape of any one child's outlook is a matter for careful assessment by the treating team, drawing together the neurology, the respiratory state, the growth and the deformity, rather than something that can be read from a curve angle or a category alone.