

Muscular Dystrophy and the Spine

A patient guide

In brief: Muscular dystrophy is a group of inherited conditions in which the muscles progressively weaken and waste because of faults in the proteins that hold muscle fibres together. The most severe and best known form is Duchenne muscular dystrophy, a condition affecting boys in which the muscle protein dystrophin is almost entirely absent; the milder Becker muscular dystrophy, in which some dystrophin is still made, and a range of other dystrophies sit alongside it. This page is written from the perspective of the spine rather than the muscle as a whole, because the spine is where one of the most predictable and important consequences of Duchenne disease plays out. As the trunk muscles weaken and the ability to walk is lost, the spine, no longer held upright by its muscular guy-ropes, tends to collapse into a long C-shaped curve known as neuromuscular scoliosis, frequently accompanied by a tilting of the pelvis. The single most important idea on this page is that in Duchenne disease the spinal deformity is tightly bound to the loss of walking and the move into a wheelchair, that long-term steroid treatment has dramatically reduced how often it develops, and that any spinal surgery must be considered against the backdrop of declining breathing and heart function.

What muscular dystrophy is, and why the spine is involved

Muscular dystrophy describes a family of genetic disorders in which muscle fibres are structurally fragile and break down faster than the body can repair them, so that over time muscle is gradually replaced by fat and fibrous tissue and strength is steadily lost. The dystrophinopathies, Duchenne and Becker, arise from faults in the gene that codes for dystrophin, a protein that acts as a shock absorber linking the inside of the muscle fibre to its surrounding scaffold. In Duchenne disease dystrophin is effectively absent and the course is severe; in Becker disease a shortened but partly functional dystrophin is still produced, so the same process unfolds more slowly and more mildly.

The reason the spine matters so much in this story is mechanical. An upright human spine is not a self-supporting column; it is held balanced over the pelvis by a continuous tension of trunk and back muscles working ceaselessly to keep it erect. When those muscles weaken symmetrically and the spine can no longer be actively balanced, it tends to give way along the path of least resistance, falling into a sideways curve and a forward or backward sag. This is the essence of neuromuscular scoliosis, and it sets the condition apart from the more familiar adolescent scoliosis of otherwise healthy teenagers, which has different causes and behaves quite differently.

The link between losing ambulation and developing scoliosis

The defining feature of the spine in Duchenne disease is how closely the development of scoliosis is tied to the loss of the ability to walk. While a boy is still walking, the act of standing and moving keeps the trunk muscles engaged and the spine extended and balanced, and significant scoliosis is uncommon. The transition into full-time wheelchair use removes that protective influence. Once a boy is seated for most of the day, the spine is no longer being actively held upright, and a curve tends to appear and then progress.

The literature describes loss of independent walking in untreated Duchenne disease as occurring around the age of nine to ten years, and the onset of scoliosis typically follows within the next couple of years, generally between the ages of ten and fourteen. The two events, becoming wheelchair-dependent and developing a spinal curve, are observed to occur together so consistently that they are often discussed as two faces of the same milestone. In the early stage the curve is frequently described as flexible and fully reducible, meaning it can still be straightened passively, but with time it stiffens and becomes a fixed, structural deformity that no longer corrects on its own.

What makes the natural history so striking is the pace. In the untreated state, neuromuscular scoliosis in Duchenne disease has historically been reported to advance rapidly once it begins, with curves increasing markedly year on year, so that a deformity that was minor on becoming wheelchair-bound could become severe within a few years. This rapid, relentless progression, occurring at a time when breathing and heart function are themselves beginning to decline, is what has placed the spine at the centre of orthopaedic thinking in this condition.

The mechanism: why a seated, weakened spine collapses

It is worth setting out plainly why the seated spine of a boy with Duchenne disease behaves the way it does. The back and trunk muscles, the long muscles running either side of the spine and the abdominal wall in front, normally act as a system of dynamic guy-ropes, constantly adjusting their pull to keep the spinal column centred over the pelvis. In Duchenne disease this whole system is weakening at once. Without active muscular balance, gravity acting on an unsupported column of vertebrae tends to drive it into a single long curve rather than the gentle natural curves of a healthy spine.

Because the muscle weakness is roughly symmetrical, the resulting deformity is characteristically a long, sweeping C-shaped curve that involves much of the spine, very different from the shorter, sharper curves of adolescent scoliosis. As the curve develops, the pelvis at its base tends to tilt out of the horizontal, a feature known as pelvic obliquity, so that one side of the seat bears far more weight than the other. This uneven loading is responsible for much of the discomfort that accompanies the deformity, as the weight of the trunk comes to rest on one buttock, and it is a major reason why correcting the deformity is concerned not only with the spine itself but with restoring a level pelvis on which a person can sit comfortably and symmetrically.

The major effect of steroid therapy on scoliosis

The most important development in this whole field over recent decades has been the recognition that long-term glucocorticoid, or steroid, treatment substantially reduces and delays the development of scoliosis in Duchenne disease. Glucocorticoids such as prednisolone and deflazacort have become a cornerstone of Duchenne care because they preserve muscle strength and function, prolong the period of independent walking, and slow the overall decline. A direct consequence of preserving strength and keeping boys walking for longer is a profound effect on the spine.

The published evidence on this point is consistent and remarkable. Comparative studies have reported that, among boys followed for many years, the proportion developing scoliosis and going on to spinal surgery was dramatically lower in those treated with glucocorticoids than in those who were not, with some long-term series describing the great majority of untreated boys requiring scoliosis surgery while only a small minority of treated boys did. Population-level data have mirrored this, showing a marked fall in the rate of scoliosis surgery for Duchenne disease over the years following the publication of the landmark studies on steroid treatment. In some treated cohorts, scoliosis severe enough to require surgery has become uncommon where it was once almost the rule.

The mechanism is thought to be largely indirect: by keeping muscle stronger and prolonging walking by a couple of years or more, steroids delay or prevent the very transition into the seated, unsupported state that drives the deformity. There may be additional effects, but the dominant theme in the literature is that the spine benefits because the loss of ambulation is postponed. This is also a reminder that steroid treatment is not without consequences of its own, including effects on bone density and on growth, which is why bone health forms part of the wider picture in these boys. The relationship described here is a pattern reported across treated populations rather than a guarantee for any individual.

Respiratory and cardiac decline, and the narrow surgical window

Running in parallel with the spinal deformity is a steady decline in the strength of the breathing muscles and in the function of the heart. In Duchenne disease the diaphragm and the muscles of the chest wall weaken over time, and the volume of air a person can move, measured as the forced vital capacity, falls progressively through the teenage

years. At the same time the heart muscle itself is affected by the same disease process, leading to a cardiomyopathy in which the pumping chambers weaken. These two trajectories, falling lung capacity and a weakening heart, are central to understanding why timing matters so much when spinal surgery is contemplated.

The difficulty is that the very deformity which surgery seeks to address tends to develop at a time when respiratory and cardiac reserve are already diminishing. A severe scoliosis can itself compress the chest and compromise breathing further, so the deformity and the respiratory decline can feed one another. This creates what is often described as a narrow surgical window: a period during which the curve has become significant enough to warrant correction, yet breathing and heart function remain robust enough to make a major operation feasible with acceptable risk. Beyond that window, as lung capacity falls below the thresholds historically used to judge fitness for surgery and as the cardiomyopathy advances, the risks of a large spinal operation climb steeply.

It is important to add that the boundaries of this window are not absolute. The literature includes accounts of successful surgery in boys with very low lung volumes when supported by intensive preoperative respiratory preparation and modern peri-operative care, and the increasing use of assisted ventilation has changed what is possible. Nonetheless, the underlying principle, that the assessment of breathing and heart function sits at the very centre of decisions about spinal surgery in Duchenne disease, remains firmly established. Decisions of this kind are made by specialist multidisciplinary teams weighing the deformity against the whole-body condition, and are entirely individual.

Why spinal fusion is the operation considered, and what it involves

When the deformity in Duchenne disease is addressed surgically, the operation considered is a long posterior spinal fusion: the placement of metal rods and anchors along the back of the spine to straighten and stabilise it, combined with bone graft so that the instrumented segment knits together into a single solid block of bone. Bracing, which is sometimes used to slow curves in other conditions, has not been found to halt the progression of neuromuscular scoliosis in Duchenne disease, which is one reason surgery has occupied such a central place once a significant curve develops.

The aims of the operation are several and worth stating clearly. The first is to stop the relentless progression of the curve, locking the spine in a corrected, balanced position. The second is to restore the ability to sit upright comfortably and symmetrically, with a level pelvis and the weight shared evenly, which has a direct bearing on comfort, on skin health over the pressure points, and on the ease of being cared for. The third, debated but supported by a number of studies, is a possible effect on the rate of respiratory decline and on survival, by preventing the deformity from further compromising the chest. The historical importance of this operation in the natural history of Duchenne disease is considerable, even as the need for it has fallen in the era of steroid treatment.

The question of fusing to the pelvis

A recurring and important consideration in Duchenne scoliosis surgery is how far down the spine the fusion should extend, and in particular whether it should stop in the lower lumbar spine or continue down to include the pelvis. This question turns on the pelvic obliquity described earlier. Because a tilted pelvis is such a prominent part of the deformity and such an important cause of uneven, uncomfortable sitting, there has long been an argument for anchoring the construct into the pelvis itself, using techniques in which rods or screws are fixed into the iliac bones, so that the pelvis can be brought level and held there.

The published experience supports the value of pelvic fixation for controlling pelvic obliquity. Series comparing fusions that stop in the lumbar spine with those carried down to the sacrum or pelvis have reported that pelvic fixation achieves and maintains a better correction of pelvic obliquity, and some studies have gone further, associating extension of the fusion to the sacrum or pelvis with improved survival. Against this, fusing to the pelvis is a larger undertaking, with its own technical demands and a recognised set of complications relating to the prominent metalwork low down and to the increased magnitude of surgery. Some authors have explored whether, in selected boys with little pelvic obliquity, the fusion can safely be stopped short of the pelvis to spare them the additional surgery. This remains an area where practice is individualised, weighing the severity of the pelvic tilt and the overall condition of the boy, and it is one of the central judgements in planning these operations.

Complications, recovery and what surgery does and does not change

A long spinal fusion in a boy with Duchenne disease is major surgery undertaken in a person whose muscles, lungs and heart are all affected by the underlying condition, and the literature is candid about the risks involved. Significant blood loss is characteristic of these long operations and is one of the factors that has been linked to outcome. Respiratory complications, including chest infections, the collapse of lung segments and the need for a period of supported breathing after surgery, are among the most important risks, which is why preoperative optimisation of the chest is given such weight. The underlying cardiomyopathy adds a further layer of anaesthetic risk, and there are reports of cardiac events during these operations attributed to the heart disease. Wound problems and issues with prominent metalwork, particularly with pelvic fixation, are also described.

It is equally important to be clear about what the operation does and does not achieve. Fusion straightens and stabilises the spine and restores level, comfortable sitting, and the evidence suggests it can slow the rate of respiratory decline and may contribute to longer survival when combined with the other elements of modern care such as assisted ventilation. What it does not do is alter the underlying muscular dystrophy, which continues to progress regardless of the state of the spine. The operation addresses one important consequence of the disease, the collapsing spine, within a condition whose overall course is determined by many factors. Recovery and the longer-term picture depend heavily on the whole-body condition, and any individual outlook is a matter for the treating team rather than something that can be read from the deformity alone.

Outlook

The outlook in muscular dystrophy as it concerns the spine has been transformed over the past few decades, and the change is best understood through the lens of steroid treatment. In the era before glucocorticoids were widely used, severe, rapidly progressive scoliosis was almost an expected part of the natural history of Duchenne disease, following hard on the loss of walking and requiring spinal fusion in a large proportion of boys. The widespread use of steroids, by preserving strength and prolonging the period of walking, has substantially reduced both the frequency and the severity of the spinal deformity, so that for many boys today significant scoliosis either never develops or develops later and more mildly.

Where a significant curve does still develop, the modern picture is one of careful, individualised judgement. The deformity is weighed against the breathing and heart function within the narrow surgical window described above, and surgery, when undertaken, aims to straighten and stabilise the spine, level the pelvis, restore comfortable sitting, and slow the decline of the chest. The decision about how far to extend the fusion, and in particular

whether to include the pelvis, is made case by case according to the pelvic tilt and the overall condition.

Across the whole field, the consistent themes are that the spinal deformity in Duchenne disease is bound up with the loss of walking, that steroid treatment has profoundly reduced it, that breathing and heart function govern the timing and safety of any operation, and that the management of the spine is only ever one part of the care of a condition that affects the whole body. None of this can be read from a category or a measurement alone; any individual situation is a matter for assessment by a specialist multidisciplinary team.