

Friedreich's Ataxia and the Spine

A patient guide

In brief: Friedreich's ataxia is an inherited, progressive disorder of the nervous system caused by a fault in a gene that supplies a protein called frataxin, and although it is best known for its effects on balance, coordination and the heart, it is also one of the most important neuromuscular causes of spinal deformity. The reason it appears on a spinal surgeon's list at all is that a sideways and rotated curvature of the spine, scoliosis, develops in the great majority of people who carry the condition, so much so that it is regarded as a near-universal orthopaedic feature rather than an occasional complication. The curve that arises in Friedreich's ataxia is not the same animal as the common adolescent idiopathic scoliosis seen in otherwise healthy teenagers; it is a neuromuscular curve, driven by the loss of the body's automatic postural control rather than by any local problem in the spine itself, and it behaves differently as a result. This page sets out, from a spinal point of view, what scoliosis in Friedreich's ataxia is, why it occurs, how the curve tends to behave over time, how it differs from idiopathic scoliosis, and what the published literature describes about the considerations that surround deformity surgery, including the heart muscle disease that so often accompanies the condition and the question of when, in the course of the illness, fusion is contemplated.

What Friedreich's ataxia is, in the briefest terms a spinal page needs

Friedreich's ataxia is the commonest of the inherited ataxias in people of European descent, and it follows an autosomal recessive pattern, meaning that two faulty copies of the gene must be inherited, one from each parent, for the condition to appear. The fault, in the great majority of cases, is an expansion of a short, repeated sequence of three genetic letters, a GAA triplet repeat, sitting within the frataxin gene on chromosome nine. This expansion silences the gene and leaves the body short of frataxin, a small protein that works inside the energy-producing machinery of cells, the mitochondria, and is especially important in tissues with high energy demands and long nerve fibres.

The result is a slow degeneration of particular pathways in the nervous system, chiefly the large sensory nerves that tell the brain where the limbs and trunk are in space, the pathways of the spinal cord that carry this information and coordinate movement, and parts of the cerebellum and its connections. The outward picture is one of progressive unsteadiness of gait and limb coordination, the ataxia from which the condition takes its name, usually beginning in childhood or adolescence, together with slurring of speech, loss of reflexes, sensory changes in the feet, and, in many people, a thickening of the heart muscle and an increased likelihood of diabetes. For the purposes of a spinal page, the salient point is not the full neurological canvas but a single downstream consequence: the loss of automatic, unconscious control of the muscles that hold the trunk upright, which is the engine of the spinal deformity that so reliably follows.

Why scoliosis is a near-universal feature

Scoliosis is so characteristic of Friedreich's ataxia that it is counted among the diagnostic features of the condition rather than treated as a chance association. The classic natural-history studies, drawing on patients followed for many years, describe scoliosis in close to the whole of a typical Friedreich's population, with figures approaching or reaching every patient in some series. It is, in short, one of the things the condition does to the body almost as a rule, and a spinal curve is frequently among the features that bring a young person to medical attention.

The reason lies in the kind of muscle weakness and imbalance the disease produces. Holding the spine straight and upright is not a passive matter of bone stacking neatly upon bone; it depends on a constant, unconscious stream of corrections by the muscles of the trunk, guided by a continuous flow of position sense travelling up from the body to the brain and back. Friedreich's ataxia attacks precisely the pathways that carry this position sense and that coordinate the postural muscles. The trunk muscles are not paralysed in the way they are in some other conditions, but the automatic system that balances them, tuning one side against the other moment by moment, is progressively lost. Deprived of this fine postural control, the growing or established spine drifts away from the midline, rotates, and settles into a curve. This is why the deformity is described as neuromuscular: its origin is in the nervous control of the trunk, not in any flaw in the vertebrae or discs themselves.

How a Friedreich's curve differs from idiopathic scoliosis

The distinction between a neuromuscular curve of this kind and the far more familiar adolescent idiopathic scoliosis is central to understanding the condition, because the two look superficially similar on an X-ray yet differ in important ways.

Adolescent idiopathic scoliosis arises in otherwise healthy young people for reasons that remain unknown, tends to produce fairly characteristic curve patterns, and exists against a backdrop of normal trunk control and normal general health. The Friedreich's curve, by contrast, is one expression of a generalised neurological disorder. Its pattern is less stereotyped. The early natural-history work noted that many Friedreich's curves do not conform to the typical shapes of idiopathic scoliosis, and that double curves involving both the thoracic and lumbar spine are common, although a proportion of patients, particularly those whose curve begins around the usual age of adolescent scoliosis, do develop curves that resemble the idiopathic type. A pronounced rounding of the upper back, a hyperkyphosis, is also frequently described alongside the sideways curve.

There are further differences that matter from a surgical standpoint. A neuromuscular curve sits within a body affected by ataxia, weakness and, often, heart muscle disease, so the person's general resilience and the safety of any intervention must be weighed quite differently than in a healthy adolescent. The trunk control that ordinarily helps to brace the spine is failing, so bracing tends to be less effective at holding these curves than it can be in idiopathic scoliosis. And because the underlying driver is a progressive disease, the natural tendency of the curve over the years cannot be read from idiopathic experience alone. These are tendencies described across the literature rather than rules that hold for every individual, but the broad lesson is consistent: a Friedreich's curve is a different problem with a different cause, and it is understood and described on its own terms.

How the curve tends to behave over time

The behaviour of scoliosis in Friedreich's ataxia has been studied since the classic series of the 1980s, and a few themes recur, while the literature is also candid about how variable the picture is from one person to the next.

A central observation is that the age at which the ataxia and the curve begin appears to bear on how the curve behaves. The natural-history work suggested that those in whom the condition and the scoliosis declare themselves early, well before the usual adolescent years, tend to develop more pronounced and more relentlessly progressive curves, whereas those whose curve appears later, around the age at which idiopathic scoliosis typically arises, may follow a gentler course more reminiscent of the idiopathic pattern. Larger curves are described as more likely to progress, and there is a recognised tendency for the curve to continue to worsen, in at least a proportion of patients,

even after skeletal growth is complete, which sets it apart from idiopathic scoliosis where progression after maturity is far less usual.

At the same time, the literature does not paint a uniform picture of inexorable collapse in every case. Some curves remain modest and relatively stable; others progress steadily and reach magnitudes at which they interfere with sitting balance, comfort and posture, particularly as the broader disease advances and walking is lost. The relationship between the severity of the neurological disease and the severity of the curve is real but loose, and the published series describe a spread of outcomes rather than a single trajectory. The honest summary is that scoliosis in Friedreich's ataxia is common, often progressive, and influenced by the age of onset and the size the curve has already reached, but that any individual course is a matter for assessment over time rather than prediction from the category alone.

Why the heart casts a long shadow over the spine

No account of the spine in Friedreich's ataxia is complete without the heart, because the two are bound together in the surgical setting in a way that is unusual among the causes of scoliosis. A thickening of the heart muscle, a hypertrophic cardiomyopathy, is part of the condition in a large proportion of patients, and over the years it may evolve, in some, towards a weakened, dilated and poorly contracting heart. Disturbances of heart rhythm also occur. Cardiac disease is, across long-term studies, the leading contributor to reduced life expectancy in Friedreich's ataxia, outweighing the neurological disability in this respect.

This matters greatly to the spine because deformity correction is major surgery. A long operation on the spine, with the blood loss, fluid shifts and physiological strain that fusion of many spinal levels entails, places real demands on the circulation, and a heart already burdened by cardiomyopathy has less reserve to meet them. The literature on Friedreich's ataxia and on neuromuscular scoliosis in general therefore treats the assessment of heart function as an integral part of thinking about spinal surgery, not an afterthought. The condition of the heart muscle, its pumping strength and its rhythm form part of the backdrop against which the risks and timing of any deformity operation are weighed, and they are among the reasons that surgery in this setting is approached with particular care and planning.

What the literature describes about deformity surgery

The published experience of scoliosis surgery in Friedreich's ataxia, while drawn from relatively small series gathered over decades rather than from large trials, points in a fairly consistent direction, and what follows describes those observations rather than offering guidance for any person.

The operation generally described for a significant, progressive Friedreich's curve is a posterior spinal fusion, in which the curved segment of the spine is straightened as far as is safe and then locked into position with metalwork, rods anchored by screws or hooks, while bone graft is laid down so that the instrumented levels gradually knit into a solid block. The aim in a neuromuscular deformity of this kind is less the cosmetic perfection sometimes sought in idiopathic scoliosis and more the achievement of a balanced, stable trunk that sits squarely over the pelvis, preserves comfortable posture and sitting balance, and resists further progression. Because neuromuscular curves are often long and sweeping and may extend low into the lumbar spine, the fusion in such cases is frequently more extensive than in idiopathic scoliosis, and decisions about how far down the spine to extend the construct, including whether to reach the pelvis, are recurring themes in the literature.

The series also document that surgery in this population carries a higher burden of complications than idiopathic scoliosis surgery, a pattern seen across neuromuscular scoliosis generally. Greater blood loss during these long operations, a higher likelihood of chest and wound problems afterwards, and the ever-present concern of the heart are all described. Set against this, the published outcomes report that fusion can reliably halt the progression of the curve, restore a more balanced posture and improve the mechanics of sitting, and the broad conclusion of the surgical literature is that deformity correction has a worthwhile role in selected patients, undertaken with careful attention to the heart and the general condition. Spinal cord monitoring during surgery, used to watch the function

of the cord while the spine is being corrected, features in this literature as well, with the recognition that the abnormal nerve pathways of Friedreich's ataxia can complicate the signals on which such monitoring relies.

The question of timing

When in the course of the illness a fusion might be contemplated is one of the more delicate questions the literature engages with, and it is shaped by the collision of two opposing pressures.

On one side, the curve tends to progress, and a larger, stiffer curve is harder to correct and is operated upon with greater difficulty and risk; on this reasoning, there is a logic to addressing a progressive curve before it becomes very severe. On the other side, the heart muscle disease that accompanies Friedreich's ataxia tends to worsen with time, and the general neurological condition declines, so deferring surgery may mean operating on a person whose cardiac reserve and overall resilience are diminished. The window in which the spine is still reasonably correctable and the heart still has adequate reserve is therefore a consideration that runs through the surgical writing on the subject. The literature describes these competing pressures and the individualised judgement they demand; it does not reduce them to a fixed age or a fixed curve size, and the published series are explicit that timing is weighed case by case, with the state of the heart given particular prominence.

Outlook

For the spine specifically, the outlook in Friedreich's ataxia turns on the magnitude and progression of the curve and on the broader course of the disease. Many curves can be controlled, and where a progressive deformity is corrected and fused, the literature describes durable straightening, a more balanced and comfortable trunk, and a halt to further progression, all of which can be of real value to a person whose mobility is in any case declining. The deformity, in short, is a manageable element of the condition.

The wider outlook, however, is governed less by the spine than by the heart and the neurological disease, which are progressive and which between them shape life expectancy and daily function. A spinal page can fairly say that scoliosis in Friedreich's ataxia is common, characteristic, often progressive, and amenable in selected patients to surgery that improves posture and arrests the curve, while also acknowledging that it sits within a serious systemic disorder whose course is determined by far more than the spine. As with every aspect of this condition, the particulars of any individual's situation, the size and trajectory of the curve, the state of the heart, the stage of the disease, are matters for careful assessment over time by the treating team rather than conclusions that can be drawn from the diagnosis alone.