

Marfan Syndrome and the Spine

A patient guide

In brief: Marfan syndrome is an inherited disorder of the body's connective tissue, caused in most cases by a fault in the *FBN1* gene that codes for a protein called fibrillin-1. Fibrillin-1 forms part of the elastic scaffolding that supports tissues throughout the body, so the condition is systemic, with well-known effects on the heart and major blood vessels, the eyes and the skeleton. This page concerns the spine specifically, because the same weakness in connective tissue that affects the aorta also shapes how the spine grows and behaves. From a spinal surgeon's point of view, Marfan syndrome tends to produce scoliosis that often appears early and can progress quickly, frequently with double or triple curves; a tendency to thoracolumbar kyphosis; and a very characteristic stretching of the lining of the spinal canal known as dural ectasia. The vertebrae themselves are often tall, slender and lightly built, with thin pedicles and loose ligaments, which alters how any deformity behaves and how it might be stabilised. Running through the whole subject is the need to weigh any spinal consideration alongside the cardiovascular and other systemic features of the condition, so that the spine is never thought about in isolation.

What Marfan syndrome is, and why connective tissue explains the spine

Marfan syndrome is a heritable disorder of connective tissue. In the great majority of people it arises from a change, or mutation, in a single gene, *FBN1*, which carries the instructions for making fibrillin-1. Fibrillin-1 is a structural protein that assembles into long fibres called microfibrils, and these microfibrils form part of the elastic framework that gives many tissues their strength, stretch and recoil. Because this framework is found throughout the body, a fault in it has consequences in many places at once, which is why Marfan syndrome is described as systemic rather than confined to any one organ.

The features most often associated with the condition reflect where elastic connective tissue matters most. The wall of the aorta, the body's main artery, depends heavily on this elastic scaffolding, which is why enlargement of the aorta is the most serious feature of the syndrome. The lens of the eye is held in place by fine connective-tissue fibres, which is why lens dislocation is characteristic. And the skeleton, including the long bones and the spine, grows under the influence of connective tissue, which is why people with Marfan syndrome are often tall and slender with long limbs and fingers.

The spine is part of this same picture. The vertebrae, the discs between them, the ligaments that bind the column together and the tough lining that surrounds the spinal cord and nerves are all connective-tissue structures. When the underlying scaffolding is altered, the spine tends to grow longer and more slender, to curve more readily, and to allow more movement at its joints. The spinal features of Marfan syndrome are therefore best understood not as separate problems but as the local expression of a body-wide difference in connective tissue.

Beyond fibrillin-1's structural role, research has shown that it also helps to regulate a signalling molecule called transforming growth factor beta, which influences how tissues grow and remodel. This helps to explain why the effects of the syndrome are not simply mechanical weakness but involve altered growth and shaping of tissues, the spine among them.

How Marfan syndrome is recognised, and where the spine fits in

The diagnosis of Marfan syndrome is made against an agreed set of criteria, most recently the revised Ghent nosology published in 2010. These criteria bring together the major systemic features, particularly enlargement of the aortic root and dislocation of the lens, the family history, the results of genetic testing for *FBN1* mutations, and a scored list of smaller, more widespread features that together build a picture of a systemic connective-tissue

disorder.

Several spinal and skeletal features contribute to this scored systemic assessment. Scoliosis or a thoracolumbar kyphosis counts towards it, as does a particular chest shape, a reduced ratio of the upper to the lower body, an increased arm span relative to height, and a number of other skeletal signs. Importantly, dural ectasia, the stretching of the lining of the spinal canal that is so characteristic of the condition, is one of the features that carries weight in this system. In this sense the spine is not a bystander in the diagnosis of Marfan syndrome but contributes several of the threads from which the overall picture is woven.

This matters for understanding the condition because it shows that the spinal features and the systemic features are intertwined. The same difference in connective tissue that places the aorta at risk is the difference that shapes the spine, and the recognition of one can prompt attention to the other. The spinal surgeon, accordingly, thinks of the spine as one window onto a whole-body condition rather than as an isolated mechanical problem.

Scoliosis in Marfan syndrome

Scoliosis, a sideways curvature of the spine usually combined with some rotation, is the most common spinal deformity in Marfan syndrome and is reported in a large proportion of those affected, with figures around three in five in many series. It is one of the most visible expressions of the condition in the spine and one of the most studied.

Several features tend to distinguish scoliosis in Marfan syndrome from the more familiar adolescent idiopathic scoliosis seen in otherwise healthy young people. It often appears earlier in childhood, sometimes in the infantile or juvenile years rather than around adolescence. The curves are frequently more complex, with double or triple patterns rather than a single curve, and they are often described as stiffer and less flexible. Crucially, those curves that are going to progress tend to do so more rapidly and more relentlessly than in idiopathic scoliosis, with the literature describing average progression of several degrees each year in untreated curves of a meaningful size.

The reasons for this pattern lie in the connective tissue itself. The same laxity of ligaments and the same altered growth that produce the tall, slender frame appear to allow the spine to curve more readily and to resist the body's natural tendency to keep itself upright and balanced. The deformity is therefore not simply a curve to be measured but the outward sign of a column that is built and held together differently.

It is worth noting that the majority of curves in Marfan syndrome remain mild and do not progress to a degree that troubles the person or calls for intervention. The literature describes a spectrum, from minor curves that are followed over time and never become a problem, through to the smaller number of larger, progressive curves that account for most of the surgical interest. This is a description of patterns observed across groups of people, not a prediction for any individual.

Why bracing behaves differently in Marfan syndrome

In adolescent idiopathic scoliosis, a back brace is a well-established way of holding a growing curve and reducing the chance that it reaches a size at which surgery is considered. In Marfan syndrome the picture is less favourable, and this difference is one of the more consistent observations in the literature.

Studies of bracing in Marfan scoliosis have reported considerably lower success rates than in idiopathic scoliosis, with one well-known series describing success in only around one in six children. Several explanations are offered. The curves often begin earlier and are more rigid, which makes them harder to control. The connective-tissue laxity may alter the way corrective pressure from a brace is transmitted through the chest wall and ribs to the spine, so that the force achieves less than it would in a stiffer, more typically built body. And the progressive forces driving the curve may simply be stronger.

The practical consequence described in the literature is that bracing in Marfan syndrome is less able to prevent a curve from reaching the range at which surgery is discussed, particularly once a curve is already of moderate size

in a child with a good deal of growth remaining. This is presented as a general observation about how the condition behaves rather than as guidance for any particular person, whose situation is always assessed individually by the treating team.

Thoracolumbar kyphosis and sagittal balance

Alongside the sideways curvature of scoliosis, Marfan syndrome has a recognised tendency to affect the spine's profile when viewed from the side, the sagittal plane. A particular pattern is thoracolumbar kyphosis, an excessive forward bend at the junction between the chest and the lower back, where the spine would normally be making a smooth transition between its natural curves.

This sagittal feature matters because the balance of the spine from front to back is as important as its straightness from side to side. A spine that is tipped too far forward at the thoracolumbar junction places the head and trunk ahead of the pelvis, and the body must work to hold itself upright against that tendency. In the context of any deformity surgery, restoring and maintaining this front-to-back balance is recognised as being especially important in Marfan syndrome, and the literature comparing it with idiopathic scoliosis notes that greater correction of sagittal balance is often required.

The same connective-tissue background that produces scoliosis underlies this kyphotic tendency. The vertebrae and discs are shaped and supported differently, and the column is less inclined to hold its natural profile. The two patterns, the sideways curve and the forward bend, frequently coexist as a kyphoscoliosis, which is part of why deformity in Marfan syndrome is often more complex than a single curve in a single plane.

Dural ectasia: the most characteristic spinal feature

Dural ectasia is among the most characteristic spinal features of Marfan syndrome and deserves particular attention because it is so closely tied to the condition. The dura is the tough outer membrane that encloses the spinal cord, the nerve roots and the cerebrospinal fluid that bathes them. In dural ectasia this membrane is stretched and ballooned outwards, most often in the lower lumbar and sacral region where the pressure of the fluid column is greatest and the surrounding connective-tissue support is weakest.

The connective-tissue basis is straightforward to understand. The dura is itself a connective-tissue structure, and when its fibrillin-rich scaffolding is altered it stretches under the steady pressure of the cerebrospinal fluid, gradually widening the dural sac and sometimes hollowing out the back of the vertebral bodies, a finding called posterior vertebral scalloping. It may also balloon out along the nerve-root sleeves to form pouches known as meningoceles. Reported prevalence varies widely with the imaging method and the threshold used, but figures from around two in five up to the large majority of those with the condition are described, which is why it is regarded as so characteristic.

Dural ectasia is usually identified on magnetic resonance imaging or computed tomography of the lower spine, where the widening of the dural sac relative to the vertebral body can be measured. It contributes to the diagnosis of Marfan syndrome as one of the scored systemic features. Many people with dural ectasia have no symptoms from it at all. Where symptoms are described in the literature, they include lower back ache, headache and other discomforts thought to relate to the altered pressure and the stretched membrane, but the relationship between the finding on a scan and any symptoms is variable. From a surgical standpoint its main significance is that a stretched, thinned dura is more fragile and that the anatomy of the lower spine is altered, both of which are relevant when any operation in that region is contemplated.

Other spinal features: spondylolisthesis, the cervical spine and protrusio

Beyond scoliosis, kyphosis and dural ectasia, several other spinal and related features are described in Marfan syndrome, each reflecting the same underlying connective-tissue difference.

Spondylolisthesis, a forward slip of one vertebra on the one below, is reported more commonly and to a greater degree in Marfan syndrome than in the general population, most often at the junction between the lowest lumbar vertebra and the sacrum. The ligamentous laxity and the altered bony architecture are thought to allow a greater slip than would usually be seen. The lower spine in the condition also shows a higher frequency of transitional vertebrae and other variations in the way the bones are formed.

The cervical spine, the neck, has its own recognised features. Radiological studies describe a tendency to focal kyphosis, increased height of the odontoid peg, basilar invagination and a degree of increased movement between the first and second cervical vertebrae, the atlas and axis, on bending forwards and backwards. This atlantoaxial laxity stems from the same ligamentous looseness that characterises the condition more generally. Frank instability requiring treatment is uncommon and reported mainly in isolated cases, but the laxity is a recognised reason for care when the neck is positioned, for instance under anaesthesia.

Protrusio acetabuli, in which the socket of the hip joint is displaced inwards towards the pelvis, is another skeletal feature of Marfan syndrome and counts towards the systemic score in the diagnostic criteria. Although it is a feature of the hip rather than the spine itself, it forms part of the same picture of altered connective-tissue-driven bony architecture in the lower trunk and is familiar to those who assess the skeleton in this condition.

The particular surgical challenges in the Marfan spine

When deformity surgery is considered in Marfan syndrome, several features combine to make it more demanding than the correction of an otherwise healthy spine, and the literature consistently draws out these differences.

The vertebrae themselves are often tall, slender and lightly built. The pedicles, the strong bony bridges through which screws are usually placed to anchor a correction, are frequently narrow and thin-walled, which makes the secure placement of fixation more exacting and means that the bony adequacy for fixation is often assessed carefully on detailed imaging beforehand. The ligaments are lax and the bone is sometimes less dense, so the anchorage of any construct must contend with tissue that holds less firmly than usual.

Dural ectasia adds its own challenge. A stretched, thinned and ballooned dura is more fragile and more easily torn during surgery, and tears of the dura with leakage of cerebrospinal fluid are reported more frequently in Marfan deformity surgery than in idiopathic scoliosis. The altered anatomy of the lower spine, including the scalloped vertebrae, further complicates work in that region.

The deformities are often larger, stiffer and more complex, with double or triple curves and a significant sagittal component, which is why the literature describes a need for longer fusions, more correction of front-to-back balance and fusion extending further down the spine than in comparable idiopathic curves. There is also a recognised tendency towards higher rates of certain complications, including greater blood loss, dural tears, problems with the fixation holding, failure of the bony fusion to consolidate and loss of correction or imbalance over time, together with a higher likelihood of further surgery. These are described as tendencies across groups of patients rather than certainties for any individual.

Coordinating the spine with cardiac and systemic risk

A defining principle in the surgical thinking around the Marfan spine is that the spine can never be considered on its own. The most serious feature of the syndrome is enlargement of the aorta, which carries a risk of dissection or rupture, and this casts a long shadow over any major surgery.

The literature is consistent that the cardiovascular and pulmonary status is evaluated carefully before any spinal deformity surgery is contemplated, because a long operation with significant blood loss and considerable swings in blood pressure places demands on a cardiovascular system that may already be vulnerable. The timing and conduct of spinal surgery are weighed against the state of the aorta and any cardiac surgery the person may have had or may need, and the assessment is shared across the disciplines involved rather than resting with the spinal team alone.

Other systemic features feed into the same coordination. The eyes, the lungs, with their tendency to spontaneous collapse, and the general connective-tissue fragility all form part of the background against which any spinal consideration is set. Medication that some people with the condition take to protect the aorta, and the broader plan of cardiovascular surveillance, are part of the picture too. The recurring theme is that the spinal features of Marfan syndrome are managed as one component of a whole-body condition, within a multidisciplinary framework, and that this coordination is itself one of the defining characteristics of how the Marfan spine is approached.

How spinal features are assessed

The assessment of the spine in Marfan syndrome rests on clinical examination and imaging, interpreted within the wider assessment of the condition as a whole.

Plain X-rays, including standing films of the whole spine and views taken bending to each side, are the standard means of measuring the size and flexibility of any curve, judging the front-to-back balance, and following whether a deformity is progressing over time. They also show features such as vertebral shape and any slip at the lower spine.

Magnetic resonance imaging, the MRI scan, is the principal way of demonstrating dural ectasia, showing the widened dural sac, any nerve-root pouches and the scalloping of the vertebrae, and it also displays the spinal cord and nerves. It is recognised as the means by which dural ectasia is identified and graded.

Computed tomography, the CT scan, gives the most detailed view of the bony architecture, including the width of the pedicles and the adequacy of the bone for any fixation, and is described as a means of planning before deformity surgery.

These investigations are not ends in themselves but tools that describe the mechanical situation of the spine and feed into the wider, multidisciplinary picture of a systemic condition. How and when they are used is a matter for the assessing team in each individual case.

Outlook

The outlook for the spine in Marfan syndrome spans a wide range, and it tracks the same spectrum that runs through the whole condition. Many people with Marfan syndrome have only mild spinal features, a small curve that is followed but never progresses, or a dural ectasia found on a scan that causes no symptoms, and for them the spine is a minor part of living with the condition.

At the other end of the range are the larger, progressive deformities that account for most of the surgical interest. Here the picture is more complex than in idiopathic scoliosis, the curves are stiffer and more likely to involve both planes, the surgery is more demanding because of the slender vertebrae, thin pedicles, lax ligaments and fragile dura, and the recognised complication and reoperation rates are higher. Even so, the literature describes that satisfactory stabilisation and correction can be achieved, and modern series report outcomes that, while harder won, can be comparable in the longer term to those in idiopathic scoliosis.

Running through the whole subject are a few consistent themes: that the spine in Marfan syndrome is the local expression of a body-wide connective-tissue difference; that scoliosis, thoracolumbar kyphosis, dural ectasia and the other features all flow from that single origin; that the slender, lax, fragile architecture shapes both the deformity and its treatment; and, above all, that the spine is always weighed alongside the cardiovascular and systemic features of the condition. Any individual outlook is a matter for careful assessment by the treating team rather than something that can be read from a category alone.