

Osteogenesis Imperfecta and the Spine

A patient guide

In brief: Osteogenesis imperfecta, often called brittle bone disease, is a group of inherited conditions in which the body makes abnormal or insufficient bone collagen, usually type I collagen, leaving the skeleton fragile and prone to fracture. It is a systemic disorder affecting the whole body, but it has particular and important consequences for the spine. The same weakness of bone that causes the long bones to break also leaves the vertebrae soft and easily compressed, so that the spinal column tends to develop characteristic problems: vertebral bodies that flatten and hollow out into a biconcave, fish-shaped form; curvatures such as scoliosis and excessive kyphosis that are common and often progressive; instability and slippage at the lower lumbar spine; and, at the other end of the column, a serious and characteristic softening at the junction between the skull and the neck known as basilar invagination. Because the bone is so fragile, any spinal surgery in osteogenesis imperfecta poses distinctive technical challenges, since screws and other metalwork must hold in bone that grips them poorly. This page describes how osteogenesis imperfecta affects the spine, what the patterns mean, and what the published evidence reports about their assessment and management, written for general understanding rather than as advice for any individual.

What osteogenesis imperfecta is, and why it affects the spine

Osteogenesis imperfecta is an inherited disorder of connective tissue, in most cases caused by a fault in one of the two genes that direct the manufacture of type I collagen, the protein that forms the flexible framework on which bone mineral is laid down. When this collagen is abnormal in quality or reduced in quantity, the bone that forms around it is weak, thin and brittle. The result is a tendency to fracture with little or no force, together with a range of other features that reflect the wide role of collagen in the body: a bluish tint to the whites of the eyes, fragile teeth, loose joints, hearing loss in adult life, and short stature in the more severe forms.

The condition varies enormously in severity. A widely used framework, originally described by Sillence, divides it into several types, ranging from a mild form compatible with a near-normal life through to forms that are severe or lethal around the time of birth. This range matters for the spine, because the more severe the underlying bone fragility, the more pronounced and the earlier the spinal problems tend to be.

The reason the spine is so often involved is straightforward. The vertebral bodies are built largely of the spongy, trabecular bone that osteogenesis imperfecta weakens most. Soft vertebrae compress and deform under the ordinary loads of standing and sitting; weak bone at the growth plates allows curves to develop and progress; and the bone of the skull base, itself softened, can gradually settle around the top of the spine. In this sense the spinal features of osteogenesis imperfecta are not separate diseases but local expressions of the same underlying fragility, and they are among the most clinically important consequences of the condition in older children and adults.

Vertebral compression fractures and the codfish vertebra

One of the most characteristic spinal findings in osteogenesis imperfecta is the deformation of the vertebral bodies themselves. Because the bone is soft, the vertebrae do not always fracture in the sudden, dramatic way a long bone does. Instead they tend to yield gradually under load, so that the vertebral body loses height and changes shape.

The classic appearance is the biconcave vertebra, in which the upper and lower surfaces of the vertebral body are pressed inwards by the cushioning discs above and below, hollowing out the bone into a shape that has long been likened to the body of a codfish. This "codfish" or "fish vertebra" pattern is so typical that it is regarded as a hallmark of the condition on a side-on X-ray. Other vertebrae flatten more uniformly, or wedge forwards, and a single spine may show several of these patterns at once. The number of deformed, biconcave vertebrae has been

used as an indicator of how severely the spine is affected, and a higher count tends to accompany a greater risk of curvature.

These vertebral changes are, in effect, compression fractures of soft bone, accumulating over years rather than occurring in a single moment. They contribute to loss of height, to a stooped posture, and importantly to the development of spinal curves, because vertebrae that are wedged or unevenly collapsed throw the column out of line. The assessment of these changes increasingly involves not only plain X-rays but also bone density measurement and formal scoring of vertebral shape, since recognising the pattern early helps to characterise the severity of spinal involvement.

Scoliosis and kyphosis: the common, progressive curvatures

Curvature of the spine is among the most frequent and troublesome spinal features of osteogenesis imperfecta. Scoliosis, a sideways curvature with rotation, is reported across a very wide range of frequencies in published series, in some cohorts affecting the majority of patients, and it becomes both more common and more severe with the more severe forms of the condition. Excessive kyphosis, an exaggerated forward rounding particularly of the thoracic spine, frequently accompanies it, producing a combined kyphoscoliosis.

The mechanism is understood to be twofold. First, the accumulation of vertebral compression fractures and the wedging and biconcave deformation of the vertebral bodies physically distort the column, so that the curve grows out of the deformed building blocks themselves. Second, the underlying weakness of the bone and the abnormal growth at the vertebral growth plates allow curves to develop and to progress in a way that ordinary spines resist. There is also growing interest in the role of disordered signalling pathways within bone, which may contribute to the malformation of vertebrae over and above the simple mechanical effect of fragility.

A particular difficulty is that these curves tend to progress, and to continue progressing into and through adult life, more relentlessly than typical adolescent curves. The progression is driven by ongoing softening and fracture rather than by growth alone, which is why curves can worsen even after skeletal maturity. This progressive tendency, combined with the fragility of the underlying bone, is what makes curvature in osteogenesis imperfecta such a demanding problem to manage.

Why bracing has a limited role

In ordinary adolescent scoliosis, a brace is a mainstay of non-operative management, used to slow or halt the progression of a curve while the spine is still growing. In osteogenesis imperfecta the situation is different, and this is an important point of contrast that the literature returns to repeatedly.

The reason is the very fragility that defines the condition. A brace works by applying corrective pressure to the trunk and the rib cage. In osteogenesis imperfecta the ribs and the underlying bone are themselves soft and prone to fracture and deformation, so the pressure a brace exerts is poorly tolerated and can deform the chest wall rather than correct the spine. The bone simply cannot provide the firm platform against which a brace needs to push. For this reason bracing is generally regarded in the published literature as ineffective at controlling curve progression in osteogenesis imperfecta, and as carrying its own risks to the fragile thorax. This stands in clear contrast to its accepted role in scoliosis arising from other causes, and it is part of why attention turns, in progressive curves, towards medical treatment of the bone and, in selected cases, towards surgery.

The craniocervical junction: basilar invagination and impression

At the very top of the spine, where the skull meets the neck, osteogenesis imperfecta produces one of its most serious and characteristic problems. The bone of the skull base is, like the rest of the skeleton, softened. Under the steady downward load of the head over many years, the soft skull base can gradually settle and the upper cervical spine, in particular the peg-like odontoid process of the second vertebra, can migrate upwards into the opening at the base of the skull. This upward settling is known as basilar invagination, or basilar impression, and a related flattening of the skull base is called platybasia.

This matters because the opening at the base of the skull, the foramen magnum, is where the brainstem and the upper spinal cord pass. As the bone settles inward and upward, it can begin to crowd these vital structures and to obstruct the normal flow of cerebrospinal fluid. Basilar invagination is reported to be present in a substantial minority of people with osteogenesis imperfecta, more so in the severe forms, and although many remain without symptoms for a long time, it can in some cases give rise to headache, neck pain, difficulty with balance and swallowing, weakness, and other signs of pressure on the brainstem and cord. Because of its potential seriousness, the craniocervical junction is regarded in the literature as a region deserving particular attention in osteogenesis imperfecta, and it is one of the most technically demanding areas to treat when intervention becomes necessary, owing to the combination of distorted anatomy and exceptionally soft bone.

Spondylolisthesis and instability of the lower spine

At the lower end of the spine, osteogenesis imperfecta is associated with a higher than usual tendency to spondylolisthesis, the forward slippage of one vertebra upon the one below, most often at the junction between the lowest lumbar vertebra and the sacrum. Studies of children and young people with the condition have reported rates of spondylolysis, a defect or stress fracture in the bony bridge of the vertebra, and of spondylolisthesis that exceed those seen in the general population.

The reasons are again rooted in the fragility of the bone. The bony bridges and pedicles that normally lock a vertebra in position can fracture, elongate or give way under load, allowing slippage to occur. There is a recognised association between slippage at the lumbosacral junction and fracture of the fifth lumbar vertebra, with one analysis reporting that the great majority of those with spondylolisthesis had sustained such a fracture and that the presence of slippage roughly doubled the likelihood of it. Unusual patterns are also described, including extreme elongation of the pedicles that allows a vertebra to slip forward while the back of the spine is left behind. These features add to the overall picture of a spine in which the structural elements that ordinarily hold the column stable are themselves weakened.

The surgical challenge: fixing metalwork into fragile bone

When spinal surgery becomes necessary in osteogenesis imperfecta, whether to correct a severe progressive curve, to stabilise a slippage, or to relieve pressure at the craniocervical junction, it presents distinctive and well-recognised difficulties that set it apart from spinal surgery in stronger bone. The central problem is mechanical: the instruments used to correct and hold the spine, principally screws placed into the pedicles, must grip bone that is soft and offers little purchase.

This poor purchase has several consequences that the literature documents. Screws may pull out of the bone, the bone around them may crumble, the metalwork may cut through or dislodge, and the correction achieved at operation may be lost over the months and years that follow. The bone may even fracture during the manoeuvres of surgery itself. To contend with this, surgeons describe a range of strategies adapted to fragile bone: spreading the load across many anchor points rather than a few, using image guidance or navigation to place screws as accurately as possible in distorted anatomy, and augmenting the grip of screws with bone cement, which is injected to fill and reinforce the soft bone around the implant so that it holds more securely. Preparing the bone medically before surgery, and timing the operation thoughtfully, are also part of the picture. Even so, the published surgical series report higher rates of implant-related complications and of loss of correction than would be expected in ordinary spinal deformity surgery, and they emphasise that operations in osteogenesis imperfecta are undertaken with these elevated risks clearly in view.

The role of bisphosphonates

A major development in the care of osteogenesis imperfecta has been the use of bisphosphonates, a family of drugs that act on bone by slowing the cells that break it down, thereby allowing bone mass to build up. Used widely in children with the condition, often given by infusion, they have been shown in the published literature to increase bone density, to reduce the rate of fractures, and to reshape and partially restore the height of collapsed vertebral bodies, an effect that is of direct relevance to the spine because it can recover some of the lost form of the vertebrae.

The evidence regarding the effect of bisphosphonates specifically on spinal curvature is more nuanced. Some studies have reported that treatment, particularly when started early and in certain types of the condition, is associated with a slower rate of scoliosis progression, while others have found no clear effect on whether a curve ultimately progresses to the point of needing surgery, an inconsistency that may reflect differences in the type of disease and the age at which treatment began. There is also an important interaction with surgery itself. Because bisphosphonates act by suppressing the natural turnover of bone, they can, in theory, slow the healing of fractures and of the cuts made in bone during an operation, and the literature accordingly discusses the timing of treatment around surgery, sometimes describing a planned pause. Bisphosphonate therapy is therefore best understood as a valuable tool that strengthens the fragile vertebrae and reduces fracture, with effects on curvature that are real but variable, and with a relationship to surgical timing that is taken into careful account.

How spinal involvement is assessed

The assessment of the spine in osteogenesis imperfecta draws on imaging and on measurement of the bone itself, building a picture of both the shape of the spine and the quality of the bone that forms it. Plain X-rays of the whole spine, taken standing where possible, are the foundation, used to measure the angle of any curve, to document the height and shape of the vertebral bodies and the characteristic biconcave deformation, and to look for slippage at the lower spine. Repeated over time, they show whether a curve is progressing.

More detailed imaging has particular value in this condition. Computed tomography gives a precise account of the bony anatomy, which is important both for understanding distorted vertebrae and, especially, for examining the craniocervical junction, where measurements of how far the upper cervical bone has settled into the skull base are used to characterise basilar invagination. Magnetic resonance imaging shows the brainstem, the spinal cord and the cerebrospinal fluid spaces, and is the means by which any compression of these structures at the skull base or elsewhere is assessed. Bone density measurement, typically by a DEXA scan, gauges the underlying fragility, though in children the result has to be interpreted with care using age- and size-adjusted scores, because the bones are small as well as weak. Together these tests describe the deformity, the bone and any effect on the neural structures, providing the information from which an individual picture is built by the treating team.

What the evidence shows about management — impersonal

It is important to state at the outset that the evidence base for managing the spine in osteogenesis imperfecta is built largely on observational studies, surgical case series, expert reviews and consensus, rather than on large randomised trials. The condition is rare, its severity varies greatly, and so the literature describes patterns and reports outcomes rather than providing fixed rules. What follows is a description of what that literature observes, not guidance for any individual, and decisions in practice are highly individualised and made within a specialist multidisciplinary team.

Medical care of the bone is described as the foundation. Bisphosphonate treatment, alongside attention to general bone health, nutrition and physical activity, is reported to strengthen the vertebrae, reduce fracture and partially restore vertebral height, and is a mainstay of management for the spine as for the rest of the skeleton.

For curvature, the literature consistently notes that bracing is of limited value because the fragile thorax tolerates it poorly, and that monitoring of curve progression over time is central. Surgery for scoliosis or kyphosis is generally described as being considered for severe and progressive curves, with reported series often referring to thresholds in the region of a substantial curve angle, balanced against the fragility of the bone and the difficulty of fixation. The surgical literature emphasises techniques adapted to weak bone, including multiple anchor points, navigation and cement augmentation, while acknowledging higher rates of implant complications and loss of correction.

For the craniocervical junction, the literature describes careful surveillance of basilar invagination, with intervention reserved for cases where there is progression or signs of pressure on the brainstem and spinal cord. The reported surgical strategies are varied and complex, including traction, decompression of the crowded region and stabilisation by fusion across the skull and upper neck, and the evidence on the best approach is acknowledged to be still debated. For spondylolisthesis, the approach described mirrors that for slippage generally, with attention to symptoms and stability.

In summary, the evidence points towards medical strengthening of the bone as the backdrop, watchful monitoring of curves and of the craniocervical junction, and surgery reserved for severe, progressive or symptomatic problems, undertaken with specialised techniques and a clear understanding of the elevated risks, all within a framework of individualised, multidisciplinary decision-making rather than fixed thresholds.

Outlook

The outlook for the spine in osteogenesis imperfecta varies as widely as the condition itself, tracking closely with its severity. In the milder forms, spinal involvement may be limited and slowly progressive, and many people lead full and active lives with monitoring and medical care of the bone. In the more severe forms, curvature and vertebral deformity tend to appear earlier, to be more pronounced, and to progress more relentlessly, and the craniocervical junction warrants particular vigilance.

Across the field, several consistent themes emerge. Medical treatment of the bone, and bisphosphonates in particular, has improved the strength of the vertebrae and reduced fracture, changing the natural history for the better. Bracing has little to offer the fragile spine. Surgery, when needed, is effective but demanding, and the fragility of the bone shapes every part of it, from the choice of technique to the rate of complications. And the craniocervical junction remains the area of greatest seriousness and the one that most rewards careful, ongoing attention. As throughout this account, any individual outlook is a matter for assessment by the treating team, taking into account the type and severity of the condition, the pattern of spinal involvement and the response to treatment, rather than something that can be read from a category alone.