

VACTERL Association

A patient guide

In brief: VACTERL association is the non-random clustering of several congenital malformations that tend to occur together more often than chance would predict, traditionally defined by the presence of at least three of six core features: vertebral defects, anal atresia, cardiac anomalies, tracheo-oesophageal fistula with or without oesophageal atresia, renal malformations, and limb abnormalities. The acronym spells out these components: V for vertebral, A for anal, C for cardiac, TE for tracheo-oesophageal, R for renal and L for limb. From the standpoint of the spine, the letter that matters most is the first one. Vertebral anomalies are among the commonest and most defining features of the association, reported in a large majority of affected individuals, and it is these congenital differences in the building blocks of the spine — hemivertebrae, butterfly vertebrae, block or fused vertebrae, and failures of segmentation — that may, in some people, give rise to congenital scoliosis. This page concentrates on the spine: what the vertebral anomalies are, how they relate to curvature, why the spinal cord itself is examined alongside the bones, and what the literature describes about assessment and surgical management of congenital curves. It is a general educational account and not advice about any individual.

What VACTERL association is, and why it is called an association

VACTERL is described as an association rather than a syndrome, and the distinction is more than semantic. A syndrome usually implies a single known cause, often a specific genetic change, producing a recognisable and consistent pattern. An association, by contrast, is a statistical observation: a group of malformations that appear together more frequently than would be expected by chance, without a single unifying cause having been established. VACTERL is the classic example of this idea in clinical medicine.

The story behind the name reflects how the picture grew. In the early 1970s, Quan and Smith described a recurring cluster they labelled VATER, standing for vertebral defects, anal atresia, tracheo-oesophageal fistula, and renal and radial limb anomalies. As further cases were studied, cardiac defects and a broader range of limb abnormalities beyond the radial side were recognised as part of the same tendency, and the acronym was expanded to VACTERL. Some authors add an S for a single umbilical artery, and overlapping labels such as VACTERL-H denote the addition of hydrocephalus.

Because there is no single laboratory test that confirms or excludes it, VACTERL association is recognised clinically, by counting the component features and, importantly, by excluding the many other conditions that can mimic the same combination. It is estimated to occur in something of the order of one in ten thousand to one in forty thousand live births. A point that runs through the literature, and that is reassuring in the present context, is that the association does not, in the great majority of affected people, impair intelligence or cognition.

The vertebral anomalies: the spinal heart of the association

Vertebral anomalies are the first letter of the acronym, and they are also among the most frequent of its features, described in a large proportion of affected individuals across published series — figures commonly cited fall in the region of sixty to ninety per cent. They are differences present from birth in the way the individual bones of the spine, the vertebrae, have formed. To understand them it helps to recall that each vertebra develops from paired blocks of embryonic tissue, the somites, which form in a rhythmic, clock-like sequence along the developing back and then condense, split and fuse in a precisely timed way to give the regular, stacked column of the mature spine.

When that orderly process is disturbed, the result is a congenital vertebral malformation. These are conventionally grouped, following classifications used in congenital scoliosis, into three broad categories. The first is a **failure

of formation**, in which part of a vertebra does not develop. A wedge vertebra represents partial failure, where one side of the body is underdeveloped; a hemivertebra represents more complete failure, where essentially only half of the vertebral body has formed, producing a triangular block that acts like a wedge driven into the column. A butterfly vertebra arises when the two halves of the vertebral body fail to fuse in the midline, leaving a cleft so that, on an X-ray taken from the front, the bone resembles a butterfly's wings.

The second category is a **failure of segmentation**, in which vertebrae that should have separated remain joined. When two or more vertebrae are fused across the front into a single block, this is a block vertebra. When the fusion runs down one side only, tethering several vertebrae along their flank, it is called a unilateral unsegmented bar. The third category is a **mixed defect**, combining elements of both, and it is this mixed pattern that often carries the most significance for the spine.

These anomalies are most often described in the thoracic and thoracolumbar regions, though they may occur anywhere. Their importance lies not in their appearance on an image in isolation but in what they may do to the balance and growth of the spinal column over time.

How vertebral anomalies lead to congenital scoliosis

Congenital scoliosis is a sideways curvature of the spine that arises from these congenital vertebral anomalies. It is fundamentally different in origin from the far commoner adolescent idiopathic scoliosis, in which the vertebrae are normally formed and the cause of the curve is unknown. In congenital scoliosis, the curve is the downstream consequence of the bones themselves having formed asymmetrically.

The mechanism is, at root, one of unbalanced growth. A healthy spine grows evenly on both sides, and the column stays straight. A hemivertebra is a wedge of bone that tilts the column to one side; if it has its own growth plates above and below, it may also continue to grow on that side while the opposite side grows normally, steadily pushing the curve onward. A unilateral unsegmented bar acts in the opposite way — it is a length of spine fused down one side, like a tether, that cannot grow on that side while the other side grows freely, again driving an asymmetric curve. The most aggressive deformities of all tend to arise when a hemivertebra and a contralateral bar occur together, so that one side is tethered and the unfused side carries an actively growing hemivertebra, producing growth that is unbalanced in both directions at once.

This is why the type and combination of anomaly, not merely its presence, govern the outlook. A balanced arrangement, in which anomalies on opposite sides cancel each other out, may produce little or no progressive curvature. A block vertebra, with no growth on either side of the fused segment, tends to behave benignly and rarely produces a large curve. It is the unbalanced patterns, and above all the unilateral bar with a contralateral hemivertebra, that the literature identifies as the most progressive.

What the natural history of congenital curves shows

The classic natural-history studies of congenital scoliosis, which followed untreated curves over years, established a clear hierarchy of progression risk according to the type of anomaly and its location. These remain among the most useful descriptions of how such curves behave.

At the benign end of the spectrum sits the block vertebra, with a very low rate of progression, often well under a degree a year, and a curve that rarely becomes severe. A single hemivertebra occupies a middle ground, with a modest and variable rate of progression. At the severe end, a unilateral unsegmented bar progresses steadily, and a unilateral bar with a contralateral hemivertebra is the most relentless pattern of all, with reported rates of deterioration that can reach the order of ten degrees or more each year during periods of rapid growth.

Location modifies these tendencies. For a given anomaly, the published series describe milder behaviour in the upper thoracic spine, more marked progression through the main thoracic region, and the most rapid deterioration at the thoracolumbar junction. The periods of fastest growth in childhood — infancy and the adolescent growth spurt — are when curves tend to advance most quickly, which is why congenital curves are watched particularly

closely across those windows. All of this describes patterns observed in groups of patients over time; it is not a prediction for any one person, in whom the behaviour of a curve is judged on its own merits.

The spinal cord, not just the spine: associated neural anomalies

A theme that recurs strongly in the literature on VACTERL and on congenital vertebral anomalies more generally is that abnormalities of the bony spine are frequently accompanied by abnormalities of the spinal cord and its coverings lying within the spinal canal. The developmental disturbance that mis-forms the vertebrae can, in the same window, affect the neural structures alongside them. This is why attention is paid to the contents of the canal and not only to the bones.

The best-recognised of these is the **tethered spinal cord**. Normally the spinal cord ends well up in the lower back and floats relatively freely; in a tethered cord it is held down at its lower end, often by an abnormally thickened or fatty terminal filament, by a fatty mass, or by other tissue, so that it is placed under abnormal tension. The reported prevalence is striking: in one frequently cited series, around two-fifths of infants with VACTERL were found to have a tethered cord requiring surgery — a figure far higher than in children with anal atresia or tracheo-oesophageal fistula in isolation. Other anomalies described in this setting include a split or duplicated cord (diastematomyelia, also termed split cord malformation), fatty lesions of the filum or cord, a fluid-filled cavity within the cord (syringomyelia or syringohydromyelia), and various forms of spinal dysraphism, in which the spine and cord have not closed completely.

Because these neural anomalies may exist without producing obvious outward signs, particularly in infancy, and because curves and operations involving the spine bring the question of the cord into sharp focus, imaging of the spinal canal forms a recognised part of the overall picture in this group.

The ribs and the chest: thoracic considerations

The spine and the rib cage develop in concert, and the same segmentation that builds the vertebrae also lays down the ribs. For this reason, congenital vertebral anomalies are frequently accompanied by rib anomalies — fused ribs, absent ribs, bifid ribs that branch, or extra ribs. Where multiple vertebral segmentation defects coincide with extensive rib fusions, the resulting picture overlaps with conditions such as spondylocostal dysostosis, which has been described in association with VACTERL.

The clinical significance of these rib and chest anomalies lies in the concept of **thoracic insufficiency**, the inability of the chest to support normal breathing or to allow the lungs to grow. A chest that is small, stiff or distorted by fused ribs and a sharply curved spine may restrict the room available for the lungs to expand and develop during the years when lung growth matters most. This consideration is one reason that, in the youngest children with extensive deformities, the goal of management is often described not simply as straightening a curve but as preserving the volume and function of the growing chest. It is also why some growth-preserving devices used in early childhood act upon the ribs as well as, or instead of, the spine itself.

How the spine is assessed in VACTERL association

Assessment of the spine in this setting rests on imaging that characterises both the bony anomalies and the neural structures within the canal, building a picture across time as the child grows.

Plain radiographs, taken from the front and the side, are the foundation. They show the vertebral anomalies — the hemivertebrae, butterfly and block vertebrae, and bars — define the shape and magnitude of any curve, demonstrate associated rib anomalies, and, repeated over time, reveal whether a curve is progressing and at what pace. Serial radiographs are the principal means of tracking congenital curves.

Magnetic resonance imaging, the MRI scan, is the modality that examines the spinal cord and the contents of the canal. It is the most sensitive way of demonstrating a tethered cord, a split cord, a syrinx or a fatty lesion, and the literature describes spinal MRI as the investigation of choice where such neural anomalies are suspected.

Because spinal ultrasound, although useful as an early screening tool in infancy, can miss a proportion of tethered cords, MRI is repeatedly highlighted as the more definitive study of the neural structures, particularly given how commonly they are involved in this association.

Computed tomography, the CT scan, with its three-dimensional reconstructions, provides the most detailed depiction of the precise bony architecture — exactly which vertebrae are malformed, how they relate to one another and to the ribs — and is especially valuable when complex anomalies are being characterised and surgery is being planned. Because VACTERL is a multi-system association, evaluation of the spine sits within a broader assessment of the heart, kidneys and other systems, since these may bear on the safety and planning of any procedure.

What the evidence shows about managing congenital curves — impersonal

It is important to be clear that the management of congenital scoliosis, including that occurring within VACTERL association, is highly individualised and rests largely on observational series, surgical case series and long-term follow-up studies rather than on large randomised trials. The descriptions that follow report what the literature observes about the strategies used, not guidance for any person.

Observation is the course described for curves judged to be at low risk of progression — balanced anomalies, block vertebrae, and small curves that are being monitored. Serial radiographic surveillance through the growing years, and particularly across the rapid-growth phases of infancy and adolescence, is the means by which a curve's behaviour is followed so that intervention, if it is ever needed, can be timed.

Bracing has a limited and particular role in congenital scoliosis. Because the curve arises from rigid, malformed bone rather than from a flexible spine, a brace cannot correct the structural component. The literature describes its use chiefly to help control flexible compensatory curves that develop above or below a rigid congenital segment, rather than to correct the congenital curve itself.

Surgical strategies are varied and are matched to the age of the child, the type and site of the anomaly, the severity and progression of the curve, and the state of the chest. **Hemivertebra excision**, the removal of the offending half-vertebra usually combined with short-segment fusion and instrumentation, is described for a well-localised, progressive curve driven by a single hemivertebra, particularly at the lumbar and lumbosacral levels, where published series report substantial correction. **In-situ fusion** and convex growth-arrest procedures aim to halt progression by stopping growth across the abnormal segment. For very young children with long, progressive curves, **growth-preserving (growing) constructs** — expandable rods anchored to the spine, and rib-based devices such as the vertical expandable prosthetic titanium rib — are described as a means of controlling the deformity while allowing the spine and, crucially, the chest to continue growing, with the aim of protecting lung development and deferring definitive fusion. Each of these approaches carries its own recognised risks and its own pattern of complications, including curve progression above or below the operated segment, junctional problems and the need for further surgery, all of which the series document.

Across this literature the consistent themes are that the type of anomaly governs the risk, that early recognition allows curves to be followed and treated before they become severe, that in the young the chest and lung growth weigh as heavily as the curve itself, and that decisions are individualised and made within a team rather than read from a category.

Outlook

The outlook in VACTERL association, viewed through the lens of the spine, is shaped above all by the type and combination of the vertebral anomalies and by whether the spinal cord and chest are involved. The single most important determinant of how a congenital curve behaves is the underlying anomaly: a balanced arrangement or a block vertebra may cause little trouble across a lifetime, whereas an unbalanced pattern, and especially a unilateral bar with a contralateral hemivertebra, carries the highest risk of a progressive and severe curve.

Set against that, several features of the broader picture are encouraging. The association does not, as a rule, affect intelligence. Many of the most serious early threats in VACTERL come from the non-spinal components — the heart, the gut and the kidneys — which are typically addressed in early life, and survival and overall outcomes have improved substantially with modern paediatric and surgical care. For the spine specifically, the value of recognising vertebral anomalies early lies in the opportunity it gives to watch curves through the growing years and to intervene, where intervention is warranted, in a planned rather than a reactive way.

What the literature underlines throughout is that this is a varied condition. Two people described under the same heading may have very different spines, very different curves, and very different journeys, and any individual outlook is a matter for assessment by the treating team rather than something that can be inferred from the label alone.