

Spina Bifida and the Spine

A patient guide

In brief: Spina bifida is a congenital condition in which the spine and the spinal cord fail to form completely during the earliest weeks of pregnancy. It belongs to a family of disorders called neural tube defects, which arise when the neural tube, the embryonic structure that becomes the brain and spinal cord, does not close as it should. The term covers a wide spectrum. At one end is spina bifida occulta, a hidden and usually harmless failure of the bony arch at the back of a vertebra to fuse, often discovered by chance and frequently causing nothing at all. At the other end is myelomeningocele, the most severe and most clinically significant form, in which the spinal cord and its coverings protrude through an opening in the back, leaving the nervous tissue exposed and damaged. From the perspective of the spine itself, the central theme is that a malformed and tethered cord sits within a malformed and often unstable spinal column, and the two influence one another throughout life. Very high rates of spinal deformity, particularly scoliosis and kyphosis, follow from the combination of paralysed and unbalanced muscles, absent or deficient bony elements at the back of the spine, and an underlying cord that may be tethered or compressed. The single most important idea is that in spina bifida the deformity of the spinal column and the disorder of the spinal cord are inseparable, and any account of the spine must hold both in view at once.

What spina bifida is, and the spectrum from occulta to myelomeningocele

Spina bifida describes an incomplete closure of the spine that occurs very early in development, when the neural tube is forming. It is helpful to picture the condition as a spectrum rather than a single entity, because the forms differ enormously in their consequences for the spine and the nervous system.

Spina bifida occulta is the mildest and commonest form. Here a vertebra, most often in the lower lumbar or sacral region, has a small gap in the bony arch that would normally close over the back of the spinal canal. The cord and its coverings remain inside the canal and the skin over the area is usually intact. In the great majority of people this is an incidental finding on an X-ray taken for another reason, causes no symptoms, and has no bearing on health. The word occulta means hidden, and that is exactly what it is. It is mentioned here mainly to set it apart from the more serious open forms, with which it shares only a name.

The closed, or occult, spinal dysraphisms are a more complex group in which the back of the spine has not formed normally but the skin is intact, sometimes with a marker such as a dimple, a tuft of hair, a fatty lump or a birthmark over the lower spine. Within this group the cord may be tethered, fixed in place by a thickened filum, a fatty mass or a bony spur, rather than hanging free as it normally would. These closed forms are distinct from myelomeningocele but are united with it by the recurring theme of a cord that is anchored rather than free.

Myelomeningocele is the severe, open form and the one that dominates the spinal surgeon's view of the condition. Here the neural tube has failed to close over a segment of the back, so that the spinal cord and the membranes around it bulge outward through a defect in the vertebrae, skin and muscle, often as a sac on the surface of the back at birth. The exposed and maldeveloped cord does not function normally below the level of the lesion, which produces weakness or paralysis, altered sensation and disturbance of bladder and bowel. It is this form that carries the very high rates of spinal deformity, the tethering, and the associated brain abnormalities described in the sections that follow.

Why the spine is so often affected

The reason spina bifida, and myelomeningocele in particular, has such a profound effect on the spine lies in the combination of problems it creates at every level of the spinal column at once.

The first is the bony deficiency itself. In an open lesion, the vertebrae at the level of the defect are split at the back, their posterior elements widely spread apart or absent, and the muscles that should attach to them are deficient or unbalanced. The spine is therefore missing part of its natural bracing at the back, the very structure that normally resists forward collapse and helps hold the column straight.

The second is the muscle imbalance that follows the cord injury. Because the cord is damaged from a certain level downward, the muscles of the trunk and around the spine are paralysed or weakened in a pattern that depends on that level. When some muscles pull and their opposing muscles do not, the growing spine is drawn progressively out of line. This is the mechanism behind neuromuscular, or paralytic, deformity, and it is why the neurological level of the lesion is so closely tied to the risk and severity of spinal curvature.

The third is the cord itself. The maldeveloped cord may be tethered at the site of the original defect or by associated abnormalities, and a tethered cord that is placed under tension as the child grows can drive or worsen deformity. Layered on top of all this, there may be congenital anomalies of the vertebrae, such as hemivertebrae or fused segments, present from birth.

Together these factors mean that the spine in myelomeningocele faces several simultaneous threats: a structurally deficient column, unbalanced forces acting upon it, an underlying cord that may tug at it, and sometimes malformed building blocks. It is the convergence of all of these, rather than any single one, that explains why spinal deformity is so common and often so severe.

Scoliosis in spina bifida: how common, and why the level matters

Scoliosis, a sideways curvature and rotation of the spine, is among the most frequent and most important spinal problems in myelomeningocele. Reported rates are high and rise with the severity and height of the lesion, with the literature describing scoliosis in a large proportion of children with myelomeningocele and in the majority of those with higher-level lesions by adolescence.

The dominant theme running through the literature is that the neurological level governs almost everything. Children with high lesions, in the thoracic region, have the greatest paralysis of the trunk muscles, the least ability to walk, and by far the highest rates of scoliosis, frequently approaching or exceeding the great majority of such children. As the lesion level descends through the lumbar segments to the sacral region, more muscle function is preserved, more children are able to walk, and the rates of scoliosis fall accordingly. Sacral-level lesions, by contrast, may be associated with relatively minor problems, although even these are not free of consequence.

The curves themselves tend to be of two broad kinds, and they often coexist. Some are congenital, arising from malformed vertebrae present from birth, and these may appear early and follow their own course dictated by the bony anomaly. Others are developmental and paralytic, arising over time from the muscle imbalance and the absent posterior bracing, and these tend to be long, sweeping curves that often extend down into the pelvis and are accompanied by pelvic obliquity, a tilting of the pelvis that affects sitting balance. Because a paralytic curve grows out of an ongoing imbalance rather than a fixed structural fault, it has a tendency to progress as the child grows, which is one reason scoliosis in myelomeningocele is watched so closely over time. These are tendencies described across groups of patients, not predictions for any individual.

Kyphosis and the congenital gibbus of myelomeningocele

Kyphosis, an excessive forward curvature of the spine, is the other major deformity of myelomeningocele, and in its severe congenital form it is one of the most striking and difficult problems in the whole of paediatric spinal surgery. It is quite distinct from the gentle, rounded kyphosis of an ordinary back.

In a proportion of children with thoracolumbar myelomeningocele, a sharp, angular kyphosis is present from birth, sometimes called a gibbus. At the apex of this deformity the spine is bent acutely forward, and because the posterior elements of the vertebrae are absent or splayed, there is nothing at the back to resist the collapse. The result can be a rigid, prominent and progressive forward angulation, often centred in the lumbar region, that

worsens as the child grows. The literature describes a natural history in which this congenital lumbar kyphosis tends to increase steadily, sometimes reaching very large angles.

The consequences of a severe kyphosis are not confined to the shape of the back. The sharp prominence at the apex stretches the overlying skin tightly over the bony point. In a child whose skin is already insensate over that region, because of the underlying neurological deficit, this combination of pressure, prominence and absent sensation is a recognised source of skin breakdown and ulceration over the apex. A large kyphosis can also compromise sitting balance, force the child into an awkward posture, crowd the contents of the abdomen and chest, and make the seating and daily care of the child more difficult. It is this combination of a rigid, progressive, skin-threatening deformity that makes the congenital kyphosis of myelomeningocele such a serious entity, and the reason that kyphectomy, the surgical removal of the apex of the deformity, has long been described in the literature for the most severe cases.

Tethered cord and progressive deformity

A tethered spinal cord is one that is abnormally fixed in position, so that instead of floating relatively freely it is anchored at its lower end or at the site of the original lesion. In spina bifida this is common, both in the open myelomeningocele, where scar tissue and the abnormal anatomy of the closed defect can fix the cord, and in the closed dysraphisms, where a thickened filum, a fatty mass or a bony spur may hold it.

The importance of tethering for the spine lies in its relationship with deformity and with neurological change. As a child grows, the spine lengthens, and a cord that is fixed at its lower end is placed under increasing tension. The literature on tethered cord describes a syndrome in which this tension is associated with a range of changes, and among these is the development or progression of spinal deformity, particularly scoliosis, alongside changes in strength, sensation, gait and bladder function. The relationship between tethering and scoliosis is one of the recurring themes of the spinal literature in spina bifida.

A good deal of attention has been given to whether releasing the tethered cord, an operation called untethering, can influence the course of an associated scoliosis. The published series describe mixed and nuanced findings. Some report that untethering can stabilise or even improve curves in selected children, particularly when the curve is still relatively small and flexible and the tethering is the principal driver. Others find that established, larger curves tend to continue to progress despite release, behaving thereafter like any other paralytic deformity. The literature therefore treats untethering not as a guaranteed remedy for scoliosis but as one factor in a complex picture, whose effect depends heavily on the size and rigidity of the curve and the wider circumstances. These are observations reported across patient groups rather than a prediction for any individual.

Congenital vertebral anomalies

Alongside the open defect itself, the spine in spina bifida frequently carries congenital anomalies of the individual vertebrae, malformations of the bony building blocks that are present from birth. These are part of the same early developmental disturbance that produced the neural tube defect.

The commonest of these are failures of formation, such as a hemivertebra, where only half of a vertebra has developed, and failures of segmentation, where two or more vertebrae are partly fused together by a bar of bone. A hemivertebra acts like a wedge inserted into the column, tilting the spine to one side, while an unsegmented bar tethers one side of the spine and prevents it from growing evenly. Both can produce a congenital scoliosis or kyphosis whose course is dictated by the underlying bony fault rather than by muscle imbalance.

What makes these anomalies important in spina bifida is that they may coexist with the paralytic deformity already described, so that a single spine can carry both a congenital curve, driven by malformed vertebrae, and a developmental paralytic curve, driven by absent bracing and unbalanced muscles. The natural history of congenital spinal deformity in the general population is itself well described, with some malformations tending to progress relentlessly and others remaining quiet, and this background informs the understanding of the bony

anomalies seen in spina bifida. The presence of these congenital elements adds to the complexity of the deformity and to the difficulty of correcting it.

Insensate skin, pressure and the neurological level

A feature of myelomeningocele that has particular relevance to the spine is the loss of sensation over the back and lower body below the level of the lesion. Because the cord is damaged, the skin in these regions is insensate, meaning it cannot feel pressure, friction or the warning discomfort that normally prompts a change of position.

This matters greatly wherever a bony prominence presses against the skin from within. A severe kyphosis, with its sharp apex, is the clearest example: the skin is stretched over a prominent bony point that it cannot feel, and the combination of unrelieved pressure and absent protective sensation is a well-recognised setting for skin breakdown and ulceration. The same principle applies to the prominences created by spinal deformity in a seated child, where pelvic obliquity and trunk imbalance concentrate pressure unevenly across the sitting surface.

The insensate skin is also a consideration in the context of surgery. Healing of the soft tissues over the spine is influenced by the abnormal skin and the often scarred, deficient tissue at the site of the original closure, and these factors form part of the backdrop to the high rates of wound problems described in the surgical literature. Across the whole subject, the insensate skin is a thread that connects the deformity, the seating, and the challenges of surgical healing, and it is one reason the spine in myelomeningocele is considered together with the soft tissues that cover it.

The surgical challenges: complications, infection and fusion to the pelvis

Surgery to correct spinal deformity in myelomeningocele is among the most demanding undertakings in spinal surgery, and the literature is consistent and candid about its difficulty. It is important to set this out plainly, because the high rates of complication are one of the defining features of the field.

Wound healing problems and deep infection are reported far more frequently after spinal fusion in myelomeningocele than after most other spinal operations. The reasons lie in the very features already described: the deficient and scarred soft tissues over the back, the insensate skin, the often poor nutritional and general health of severely affected children, the presence of a ventricular shunt, and contamination from a neuropathic bladder and bowel. Studies of deep wound infection after neuromuscular scoliosis surgery, of which myelomeningocele is one of the highest-risk groups, document substantial infection rates and explore the factors that contribute to them.

Beyond infection, the literature describes high rates of other problems: failure of the bone to fuse, known as pseudarthrosis; loosening, breakage or prominence of the metalwork used to hold the correction; and the need for further surgery. The deficient posterior bony elements give the surgeon less native bone to fuse to and fewer secure points of fixation, which compounds the mechanical difficulty.

A recurring and characteristic feature is the frequent need to extend the fusion down to the pelvis. Because the long paralytic curves of myelomeningocele so often involve pelvic obliquity and extend into the sacrum, achieving a balanced, level pelvis for sitting frequently requires anchoring the lower end of the construct into the pelvis itself. Fixation to the pelvis is mechanically demanding and adds its own set of considerations and risks. Taken together, the high complication and infection rates, the difficulty of obtaining solid fusion, and the frequent need for pelvic fixation make spinal surgery in myelomeningocele a field in which the balance of benefit and risk is weighed with particular care.

The Chiari II malformation, hydrocephalus and the shunt

Almost all children with myelomeningocele have an associated abnormality at the base of the brain called the Chiari II malformation, in which the lower part of the brain and brainstem are displaced downward through the opening at the base of the skull into the upper spinal canal. This is closely linked to hydrocephalus, a build-up of fluid within the brain, which very commonly requires a shunt, a tube that drains the fluid away, usually placed in

early life.

These features may seem to belong to the brain rather than the spine, but they interact with the spinal picture in ways that matter. The Chiari II malformation involves the very top of the spinal canal and the junction between the brain and the cord, and changes here can produce or contribute to neurological symptoms. Hydrocephalus and the function of the shunt are part of the wider neurological balance of the child; a shunt that is not working well can cause a change in neurological state, which in turn may be relevant to the cord and the spine.

For the spinal surgeon, the presence of a shunt is also a practical consideration in its own right, both as a potential source of infection and as part of the complex medical background that any spinal operation must take into account. A change in a child's neurological condition or in the behaviour of an established scoliosis may prompt consideration of several interrelated causes at once, including the shunt, the Chiari malformation and tethering of the cord. This is one further illustration of the central theme of spina bifida and the spine: that the brain, the cord and the column are bound together, and none can be considered fully in isolation from the others.

Outlook

The outlook for the spine in spina bifida varies as widely as the condition itself, and it tracks closely with the form of the condition and, in the open lesions, with the neurological level.

For spina bifida occulta and many of the milder forms, there is often no consequence for the spine at all, and the finding carries no particular implication for spinal health. For myelomeningocele the picture is more involved, and the height of the lesion is the strongest guide: higher lesions carry greater paralysis, lower rates of walking, and far higher rates of severe scoliosis and kyphosis, while lower and sacral lesions are associated with less severe spinal problems. The deformities themselves, where they occur, tend to develop and progress over the years of growth, which is why the spine is followed over time.

Where surgery is undertaken, the literature describes both its considerable value in restoring sitting balance, controlling a progressive deformity and addressing a threatening kyphosis, and its considerable risks, with high rates of infection, wound problems and the need for further surgery. Across the whole field the consistent themes are that the deformity of the column and the disorder of the cord are inseparable, that the neurological level shapes the severity of nearly everything, that the skin and soft tissues are a constant consideration, and that any individual outlook is a matter for assessment by the treating team rather than something that can be read from a category or a measurement alone.