

Spina Bifida Occulta

A patient guide

In brief: Spina bifida occulta is a very common, minor anatomical variant of the spine in which one of the bony arches at the back of a vertebra, most often the lowest lumbar vertebra (L5) or the top of the sacrum (S1), fails to fuse completely in the midline during early development. The word "occulta" means hidden, and that single word captures almost everything that matters about it. The defect is purely in the bone; the spinal cord, the nerves and their protective coverings, the meninges, are entirely normal and remain safely enclosed within the spinal canal. Nothing is exposed and nothing protrudes. For this reason spina bifida occulta is, in the overwhelming majority of people, completely harmless and causes no symptoms whatsoever. It is so common that it is found in roughly one in ten to one in five of the population, and it is usually discovered entirely by chance on an X-ray or scan performed for some other reason. It is fundamentally different from the serious open forms of spina bifida, the meningocele and the myelomeningocele, in which neural tissue or its coverings are involved. The single most important idea is that occulta means hidden and closed: a quiet variation in the shape of a bone, not a disease of the nervous system.

What spina bifida occulta is (and why "occulta" means hidden and harmless)

Each vertebra in the spine is formed from a block of bone at the front, the vertebral body, and a ring of bone at the back, the neural arch, which arches backwards to enclose and protect the spinal cord and nerves. During development in the womb, this posterior arch grows from each side and fuses together in the midline, eventually meeting to complete the bony ring and form the spinous process, the knobble that can be felt running down the centre of the back. Spina bifida occulta is simply a failure of that midline fusion to complete at one or more levels, leaving a small gap in the bony arch where the two halves did not quite meet.

The word "occulta" is Latin for hidden, and it is the key to the whole subject. The defect lies in the bone alone, concealed beneath intact skin and muscle, with no outward sign in most people and nothing visible on the surface. Crucially, the contents of the spinal canal, the spinal cord, the nerve roots and the meninges that wrap around them, are all completely normal and remain fully enclosed. There is no exposure of neural tissue, no protruding sac, and no opening to the outside world. This is what is meant by a "closed" defect, and it is the reason the condition is, in the vast majority of cases, of no consequence at all.

It is helpful to think of spina bifida occulta less as a disease and more as an anatomical variant, a normal variation in the way a bone has formed, in the same broad category as the small differences between one person's skeleton and another's. A person with this finding on an X-ray is generally expected to have no physical limitations, no muscle weakness and no increased tendency to develop spinal problems because of it.

How common it is

Spina bifida occulta is extremely common, and is by a wide margin the most frequent of all the forms of spina bifida. Estimates of how often it occurs vary considerably depending on the population studied and on the imaging method used, but figures of roughly ten to twenty per cent of the general population are widely quoted, with one commonly cited estimate around twelve per cent.

The reported range is in fact wider still. Large studies of normal adults have found abnormalities of posterior arch formation in over a fifth of people, and some authorities have suggested that if the back of the spine were imaged carefully in every healthy adult, as many as one in three might be found to have some degree of incomplete fusion. The variation between studies reflects how the finding is defined, which vertebrae are examined and how closely

they are scrutinised, rather than any real disagreement about the basic point: this is a very common variant indeed.

A few patterns are consistently described. The defect is most often found at the junction between the lumbar spine and the sacrum, particularly at the fifth lumbar vertebra and the first sacral segment, and it is reported somewhat more frequently in men than in women. It is a feature of how the spine formed in early life and, because the bony arch does not fuse spontaneously once development is complete, it persists unchanged into adult life rather than appearing or disappearing over time.

Why it differs fundamentally from open spina bifida

The phrase "spina bifida" covers a spectrum that runs from the trivial to the serious, and it is essential to distinguish where on that spectrum the occulta form sits. Doctors divide the conditions into closed (occult) and open (aperta) forms, and the difference between them is profound.

****Spina bifida occulta is a closed defect.**** Only the bony arch is affected. The skin over it is intact, nothing protrudes, and the spinal cord, nerves and meninges are normal and fully enclosed. This is the mildest end of the spectrum and is, in most people, harmless.

****Meningocele**** is one of the open, or aperta, forms. Here the bony defect is larger, and the meninges, the membranes that surround the spinal cord, push outward through the gap to form a fluid-filled sac on the back. The spinal cord and nerves themselves are usually not contained within the sac, which is why meningocele tends to be less severe than the form described next, though it is still a significant condition requiring specialist care. It is the least common of the three main types.

****Myelomeningocele**** is the most severe form of spina bifida. In this condition the sac that protrudes through the defect contains not only the meninges and cerebrospinal fluid but also the spinal cord and nerves themselves, which are exposed and abnormally developed. This is the form most people picture when they hear the term spina bifida, and it is associated with substantial neurological consequences, often affecting movement in the legs, bladder and bowel control, and sometimes the brain.

The gulf between these is enormous. Spina bifida occulta shares only a name and a broad developmental origin with the open forms; in every way that matters to a person's health it is a different entity. Naming alone causes a great deal of unnecessary alarm, because the words "spina bifida" carry associations with the severe open conditions that simply do not apply to the harmless closed variant.

Why it happens

Spina bifida occulta arises very early in development, during the period in which the neural tube, the structure that becomes the brain and spinal cord, closes and the surrounding bony spine forms. In the occulta variant, the neural tube itself closes normally, and it is only the later fusion of the bony arches behind it that is left incomplete. This is why the nervous system is structurally normal in this form, unlike the open conditions in which the closure of the neural tube itself is affected.

The precise causes are not fully understood and are thought to involve a combination of genetic, nutritional and environmental influences acting on the developing spine. Folate, a B vitamin, is well established as important in the proper closure of the neural tube, and adequate folate intake around the time of conception is recognised as reducing the risk of the serious open neural tube defects. For the minor occulta variant specifically, the picture is less clear-cut, and many people who have it have no family history of spina bifida and are not found to be at any meaningfully increased risk of having a child with the severe forms.

For the great majority of people, no cause is ever identified and none needs to be sought, because the finding is simply a common variation in how the bony spine happened to form.

Usually an incidental finding

Most people who have spina bifida occulta never know it, and for good reason: it usually produces no symptoms at all and gives no outward sign of its presence. It is therefore most commonly discovered entirely by accident, when an X-ray, CT or MRI scan of the lower back is performed for some unrelated reason, such as the investigation of back pain, an injury or another complaint, and the radiologist happens to note the incomplete fusion of an arch in passing.

This incidental nature is one of the reasons the finding can cause alarm out of proportion to its significance. Seeing the words "spina bifida" written in a scan report is understandably unsettling, even though, in the context of the occulta variant, it describes a minor and usually meaningless quirk of the bony anatomy. The fact that it was found while looking for something else, rather than because it was causing trouble, is itself a strong clue to how little it generally matters.

Because it is so often silent, there is rarely any reason to look for spina bifida occulta deliberately, and it is not something that is routinely screened for. It tends to come to light only because the lower spine has been imaged for another purpose.

Skin markers as occasional pointers

While spina bifida occulta itself usually leaves no trace on the surface, a minority of people have a small mark on the skin of the lower back overlying the site of the bony defect. The marks most often described are a tuft or patch of hair, a small dimple or pit, a birthmark or pigmented patch, a fatty lump beneath the skin, or a small reddish mark. These are collectively known as cutaneous stigmata, the visible skin signs that can sometimes accompany a defect in the underlying spine.

In most instances these marks are themselves entirely innocent and are of no greater significance than the bony variant they sit above. Their importance lies in a different and more subtle role: occasionally, particularly when there is more than one such mark, or when a mark is unusual in being large, deep, off to one side of the midline or accompanied by others, they can serve as an outward pointer to a less common and more significant underlying condition known as occult spinal dysraphism. In other words, the skin sign is sometimes a flag that prompts a closer look, even though the simple bony variant on its own is harmless.

This is the reason that, in infants and small children, the discovery of certain skin markers over the lower spine may lead to imaging, usually an ultrasound scan in a newborn or an MRI scan in an older child, to look beneath the surface. A single, small, simple midline dimple close to the buttock cleft, with no other features, is generally not associated with any underlying problem and does not by itself prompt imaging; it is the atypical or multiple markers that draw attention.

The rare association with occult spinal dysraphism and tethered cord

Although simple spina bifida occulta is harmless, it sits at the very mildest end of a broader family of closed spinal conditions, and it is important to understand the rare but real connection between them. The broader group is called occult spinal dysraphism, an umbrella term for a range of hidden, closed abnormalities in which the spinal cord or its surrounding structures are subtly malformed beneath intact skin. These include such things as a fatty growth attached to the cord, a band of fibrous tissue, a dermal sinus tract, a thickened or tight filum terminale (the thread of tissue at the lower end of the cord), or a split in the cord.

The most clinically relevant of these conditions is the tethered cord. Normally the spinal cord hangs relatively freely within the spinal canal and is able to move a little. In a tethered cord, the lower end of the cord is abnormally fixed or anchored, often by one of the structures just described, so that as a child grows the cord is stretched rather than able to ride up freely. This stretching can, over time, interfere with the function of the lower cord and nerves. Spina bifida occulta of the bony arch is frequently present alongside these conditions, which is why it can act as a signpost, even though the bony defect itself is not what causes the trouble.

When a tethered cord or other dysraphism does produce symptoms, they tend to involve the lower limbs, the bladder and bowel: examples include leg weakness or numbness, changes in the shape of a foot or leg, difficulties with walking, problems with bladder or bowel control, recurrent urinary infections, back or leg pain, and sometimes a curvature of the spine. These features may appear at any age from infancy to adulthood. It must be stressed that these associated conditions are uncommon, and that the great majority of people with simple spina bifida occulta have none of them. The bony variant alone, without any of these deeper abnormalities, is not expected to cause such problems.

What the evidence shows about symptoms and back pain

A question that frequently arises is whether spina bifida occulta is responsible for back pain. The weight of evidence indicates that simple spina bifida occulta is generally not a cause of back pain. Systematic reviews of imaging studies have found no consistent relationship between the presence of the bony variant and the occurrence of back pain. Because the variant is so common in the general population, it is often present purely by coincidence in people who happen to have back pain arising from quite separate and ordinary causes, and its appearance on a scan can be a red herring that distracts from the true source of the trouble.

The literature is not entirely uniform, and a small number of studies have reported associations, for example a suggestion that back pain might be more troublesome in some people who also have the variant, or that it coexists with other minor structural variations such as a transitional vertebra or with spondylolysis, a small stress defect in part of the vertebral arch. These reported associations are inconsistent and do not establish that the occulta variant itself is the cause. The prevailing view is that finding spina bifida occulta on a scan, in a person with back pain, does not by itself explain that pain, and that other, more usual causes are generally the relevant ones.

This is a description of what studies of groups of people have found, not a statement about any individual, and it remains the case that the relationship between what is seen on a scan and what a person experiences is, at most, loose.

How it relates to other minor spinal variants

Spina bifida occulta is one of several common variations in the way the lower spine forms, and it is sometimes found alongside others. One that is occasionally mentioned in the same breath is spondylolysis, a small defect in a part of the vertebral arch called the pars interarticularis, which is reported more often in people who also have spina bifida occulta. The reasoning offered is that the slightly altered shape of the arch may place a little more stress on this region. Another is the lumbosacral transitional vertebra, a variant in which the bottom lumbar vertebra takes on some features of the sacrum, or vice versa.

The general theme running through all of these is the same: they are common quirks of how the bony spine developed, frequently found together, often discovered by chance, and in most people of limited or no consequence. Recognising spina bifida occulta as a member of this family of minor variants helps to keep it in perspective, as a description of a shape rather than a diagnosis of a disease.

What it usually means in practice

For the overwhelming majority of people in whom it is found, spina bifida occulta means nothing of practical importance and requires nothing to be done about it. It is a harmless, hidden variation in the bony anatomy of the spine, discovered by chance, and not in itself a source of symptoms, limitation or future risk. The literature is consistent that a person with this finding alone is expected to lead an entirely normal life, unaffected by it.

The exceptions, which are uncommon, are those situations in which the finding sits alongside other features that suggest one of the rarer closed conditions: in particular, certain skin markers over the lower spine, or symptoms pointing to the lower spinal cord and nerves, such as leg, bladder or bowel disturbances. It is the presence of these additional features, rather than the bony variant itself, that may lead to further assessment, usually beginning with imaging to look beneath the surface. In their absence, the variant is generally left undisturbed and unremarked.

The reassuring message that runs through the whole subject is captured by the word "occulta": hidden, closed and, for almost everyone, harmless.

Outlook

The outlook for a person found to have spina bifida occulta is, in the great majority of cases, entirely reassuring. The variant is lifelong, in the sense that the bony arch does not fuse later, but it is also stable and silent, and it does not progress, deteriorate or give rise to new problems over time. People with this finding alone are expected to have normal strength, normal sensation, normal walking and no particular vulnerability to spinal disease as a consequence of it.

Where the picture is different, it is not the bony variant that determines the outlook but the rarer associated conditions, the tethered cord and the other forms of occult spinal dysraphism, that occasionally accompany it. In those uncommon situations the outlook depends on the specific abnormality, on whether and how it produces symptoms, and on the assessment of a specialist team, and it is a quite separate matter from the simple incidental finding.

Across the whole subject, the consistent themes are that spina bifida occulta is one of the commonest of all spinal variants, that "occulta" means hidden and closed and harmless, that it is fundamentally different from the serious open forms of spina bifida, that it is almost always an incidental and meaningless finding, and that the great majority of people who have it need nothing done about it at all.