

Down Syndrome (Trisomy 21) and the Spine

A patient guide

In brief: Down syndrome, or trisomy 21, is the most common chromosomal condition in humans, arising when a person carries an extra copy of chromosome 21. It is associated with a recognisable pattern of features across many body systems, but from the standpoint of spinal care the central theme is a generalised laxity of the ligaments and a softness of muscle tone that together loosen the joints throughout the skeleton, and most importantly at the top of the neck. The junction between the skull and the first two vertebrae is the area of greatest concern. Here, loose ligaments and, in some individuals, an underdeveloped or separated peg of bone at the front of the second vertebra can allow more movement than the spinal cord can safely tolerate. This is termed craniocervical instability, of which atlantoaxial instability is the best known form. Further down the spine, the same laxity contributes to a raised rate of scoliosis and of slippage of one vertebra on another. The single most important idea is that the great majority of this instability is detected on imaging without ever causing symptoms, and that the small minority in whom the spinal cord is genuinely at risk is what spinal assessment is designed to identify.

What Down syndrome is, and why it affects the spine

Down syndrome results from the presence of an additional copy, in whole or in part, of chromosome 21. In roughly ninety-five per cent of cases the extra chromosome is present in every cell, a pattern called trisomy 21; the remainder arise through translocation or mosaicism. It is the commonest chromosomal disorder, occurring in the region of one in seven hundred to one in a thousand live births, with the likelihood rising with maternal age. Survival and life expectancy have improved markedly over recent decades, so that the condition is now very much one of adult as well as childhood medicine, and the spinal questions it raises follow people across the whole of life.

The reason Down syndrome has such a consistent effect on the spine lies in the connective tissues. Chromosome 21 carries the genes for type VI collagen, a protein woven through ligaments, muscle, bone and cartilage throughout the body. The extra copy of the chromosome is thought to alter the way this collagen is produced, and the result is a generalised looseness of the ligaments together with low muscle tone, the combination often described as hypotonia. Ligaments are the strong bands that hold one bone to the next and limit how far a joint can move; when they are lax, joints across the body become more mobile than usual. In the spine this laxity is felt most acutely at the very top of the neck, where the margin between safe and unsafe movement is narrowest, but it also shows itself lower down as scoliosis and as slippage between vertebrae.

The craniocervical junction: where the concern is greatest

The region where the skull meets the spine is known as the craniocervical, or craniovertebral, junction. It is made up of the base of the skull, the first cervical vertebra, the atlas, and the second, the axis. The atlas is a ring of bone that cradles the skull and allows the head to nod. The axis carries an upward projection of bone at its front, the odontoid process or dens, around which the atlas and the head rotate. A strong band, the transverse ligament, holds this peg snugly against the back of the front arch of the atlas, and it is this arrangement that keeps the spinal cord safe while allowing the head to turn.

In Down syndrome this elegant mechanism is vulnerable in two ways. First, the transverse ligament and the surrounding ligaments are often lax, allowing the atlas to slide forward on the axis more than it should. Second, in a proportion of individuals the bony peg itself is abnormal, being underdeveloped or, in the condition known as os odontoideum, separated into a free-floating ossicle. Either change reduces the joint's ability to hold its alignment as the neck moves. The space through which the spinal cord passes can then be encroached upon, particularly when the neck is bent forward. This is the anatomical basis of craniocervical instability in Down syndrome, and it explains why the upper neck receives such particular attention.

Atlantoaxial and atlanto-occipital instability

Craniocervical instability in Down syndrome is conventionally divided into two patterns. Atlantoaxial instability describes excessive movement between the atlas and the axis, the first and second vertebrae. Atlanto-occipital instability, sometimes called occipito-axial or atlanto-occipital instability, describes excessive movement between the skull and the atlas. The two can occur separately or together, and both arise from the same background of ligamentous laxity, sometimes compounded by abnormalities of the bones of the region such as underdeveloped occipital condyles or articular facets that reduce the natural locking together of the joint surfaces.

Atlantoaxial instability is the more familiar of the two and has been studied for decades. Reported figures vary, but radiographic atlantoaxial instability is described in something of the order of ten to twenty per cent of people with Down syndrome, with some series quoting ranges as wide as six to thirty per cent depending on how it is defined and measured. Atlanto-occipital instability is reported in a similar proportion, with one adult series placing it at around seventeen per cent. The wide spread of these figures reflects differences in the populations studied and, above all, differences in the radiographic thresholds chosen, a theme that runs through the whole subject.

The atlanto-dental interval and how instability is measured

The principal measurement used to describe atlantoaxial instability is the atlanto-dental interval, sometimes called the atlas-dens interval and abbreviated to ADI. It is the gap, measured on a side-on radiograph, between the back of the front arch of the atlas and the front of the odontoid peg. When the ligaments are intact and tight this gap is small; as they loosen and the atlas drifts forward, the gap widens. In adults an interval greater than around three millimetres is generally regarded as beyond the normal range, whereas in children somewhat larger values, up to four or five millimetres, may fall within normal limits because of the looser ligaments of childhood.

The atlanto-dental interval, however, tells only part of the story. Just as important, and arguably more so, is the space left for the spinal cord itself, often called the space available for the cord. As the atlas slides forward, the peg of bone occupies more of the canal and the room for the cord shrinks. For this reason the space available for the cord is widely regarded as the measurement that correlates most closely with actual risk to the cord, and a narrowing of this space below a critical figure is treated with more concern than a widened atlanto-dental interval alone. Measurements taken with the neck bent forward and backward, the flexion and extension views, are used to reveal how much the alignment changes through the range of movement, since the danger lies in the abnormal motion rather than in any single static position.

Asymptomatic versus symptomatic instability

The most important distinction in this whole field is between instability that is merely visible on imaging and instability that is actually causing harm to the spinal cord. The two are very far from the same thing. The great majority of people with Down syndrome who have a widened atlanto-dental interval have no symptoms whatever and never develop any. Asymptomatic radiographic instability is common; symptomatic instability, in which the spinal cord is genuinely compromised, is uncommon, with figures often quoted in the region of one to two per cent of people with Down syndrome.

This gulf between what is seen and what is felt is the reason that a measurement on a scan, taken in isolation, is a poor guide to risk for any individual. A widened interval indicates that the joint is loose, but looseness alone does not mean the cord is in danger. What raises genuine concern is the combination of marked radiographic instability, a narrowed space for the cord, and, above all, symptoms or signs that the cord is being affected. The features that point to spinal cord involvement, collectively termed myelopathy, include a change in the way a person walks, increasing clumsiness of the hands, a deterioration in bladder or bowel control, new neck pain or a tilting of the head, and on examination the brisk reflexes and other signs that indicate pressure on the cord. It is the appearance or progression of such features, rather than a number on a radiograph, that marks the small group in whom the instability has become clinically meaningful.

Os odontoideum and bony abnormalities of the peg

Among the bony abnormalities that contribute to instability in Down syndrome, the most significant is os odontoideum. In this condition the odontoid peg, instead of being a single continuous projection of bone fused to the body of the axis, is separated into a rounded ossicle that sits above a gap. The transverse ligament can then no longer anchor a solid peg, and the upper segment is free to move with the atlas, producing instability that may be considerable. Underdevelopment of the peg, termed odontoid hypoplasia, has a similar effect to a lesser degree.

Os odontoideum is important because it combines, in a single individual, the ligamentous laxity of Down syndrome with a structural failure of the very bone that the ligaments are meant to restrain. The result can be instability that is both greater in degree and more likely to threaten the cord, and it is among the situations in which the literature most consistently describes a role for surgical stabilisation. Because the bony abnormality is fixed, whereas ligamentous laxity may be partly compensated, the presence of os odontoideum tends to weigh heavily in any assessment of the upper neck.

Scoliosis and the rest of the spine

Although the craniocervical junction dominates attention, the same ligamentous laxity that affects the upper neck has effects along the length of the spine. Scoliosis, a sideways curvature of the spine often combined with a rotation of the vertebrae, occurs more often in people with Down syndrome than in the general population, with prevalence figures commonly cited in the region of around five to nine per cent. The curves seen in Down syndrome are generally regarded as a form of syndromic scoliosis, distinct from the common adolescent idiopathic form, and they arise against the background of low muscle tone and loose ligaments that allows the growing spine to drift out of line.

The same laxity contributes to spondylolisthesis, in which one vertebra slips forward on the one below, most often in the lower lumbar spine. Taken together, the spinal pathology of Down syndrome has been described as falling into three broad areas: instability of the occipitocervical junction at the top, scoliosis through the trunk, and spondylolisthesis at the base. Beyond the spine itself the connective-tissue laxity expresses itself in hip and kneecap instability, flat feet and other features, all reflecting the same underlying softness of the supporting tissues.

How the spine is assessed

Assessment of the spine in Down syndrome rests on a combination of careful clinical evaluation and imaging, with the two weighed together rather than either taken in isolation. Clinical assessment looks for the symptoms and signs of pressure on the spinal cord: changes in walking and balance, in hand function, in bladder and bowel control, the presence of neck pain or an abnormal head position, and on examination the reflexes and other neurological signs that indicate myelopathy. The emphasis in current practice has shifted firmly towards this clinical vigilance, with major paediatric bodies no longer recommending routine screening radiographs of the neck in children who have no symptoms, on the grounds that a radiographic measurement in an asymptomatic child predicts future trouble poorly.

Plain radiographs, including the side-on flexion and extension views, remain the standard means of measuring the atlanto-dental interval and the space available for the cord and of demonstrating how the alignment changes through movement. **Magnetic resonance imaging, the MRI scan**, shows the spinal cord directly, reveals whether it is being compressed and whether it has sustained any change in its substance, and can be performed in flexed and extended positions to capture dynamic narrowing that a single neutral image would miss. **Computed tomography, the CT scan**, displays the bony anatomy of the region in fine detail and is the clearest means of demonstrating an os odontoideum or other abnormality of the peg. The space available for the cord and the atlanto-dental interval are the measurements that have been shown to correlate most closely with actual cord compression, and it is the dynamic studies, capturing movement, that carry the greatest weight.

What the evidence shows about management

It is important to state at the outset that the evidence guiding the management of craniocervical instability in Down syndrome is built largely on retrospective series, case reports and systematic reviews of these, rather than on large randomised trials, and that there are no universally agreed numerical thresholds at which intervention becomes necessary. Systematic reviews of surgical treatment in children with Down syndrome have explicitly noted the sparseness and inconsistency of the data. The descriptions that follow report what the literature observes, not guidance for any individual, and decisions in practice are made on a case-by-case basis by the treating team.

Observation is the course described for the large majority, in whom instability is asymptomatic and the spinal cord is not compromised. Here the literature describes periodic clinical review, with attention to the emergence of any symptoms or signs of cord involvement, rather than intervention. **Activity considerations** feature in the literature, with certain high-risk activities, particularly those involving forced bending of the neck or the risk of collision, historically restricted for individuals with demonstrated instability; the requirements of organised sport in this regard have themselves evolved over time. **Care around anaesthesia** is a recurring theme, because the positioning of the neck during the induction of anaesthesia and during airway management carries a theoretical risk in a person with a lax upper neck, and the literature describes meticulous attention to neck position in this setting.

Surgical stabilisation is reserved in the literature for the minority in whom instability is symptomatic, with established or progressive myelopathy, marked radiographic instability with cord compromise, or a structural lesion such as os odontoideum threatening the cord. The operation joins the unstable segments into a single solid block of bone, a fusion, most often across the atlas and axis or, where the skull itself is involved, from the occiput down onto the upper cervical spine. Techniques described include screw-and-rod constructs such as the Goel-Harms fixation and occipitocervical instrumentation. The literature is candid that these operations are demanding, particularly in young children with small and fragile bones, and that complications including failure of the bones to fuse, infection and hardware problems are reported more often than in routine spinal surgery, a reflection of the low bone density, the high ratio of head to body size and the other features of the condition. The threshold for surgery is therefore weighed carefully against its risks.

Scoliosis management in this context

The management of scoliosis in Down syndrome follows the broad principles applied to syndromic scoliosis generally, with observation of smaller curves, the consideration of bracing in some growing spines, and surgical correction by instrumented fusion reserved for larger or progressive curves. The literature consistently records, however, that surgical correction in Down syndrome carries a higher rate of complications than the same surgery for adolescent idiopathic scoliosis, with wound healing problems and infection featuring particularly prominently and overall complication rates in some series exceeding half of those operated upon. These observations describe the experience of groups of patients and the particular care that the condition demands, rather than any prediction for an individual.

Outlook

The outlook for the spine in Down syndrome is, for most people, reassuring, and it follows closely the distinction that runs through this whole account: between instability that is merely visible and instability that genuinely threatens the spinal cord. The overwhelming majority of those with radiographic atlantoaxial instability remain free of symptoms throughout life, and the loose upper neck of Down syndrome, though common, only rarely translates into harm to the cord. The role of assessment is to identify, from within this large group, the small minority in whom symptoms or signs of myelopathy, marked instability with cord compromise, or a structural lesion such as os odontoideum indicate that the cord is genuinely at risk.

Where the cord is not at risk, the literature describes watchful clinical follow-up rather than intervention. Where it is, stabilisation can protect the cord, although the surgery is demanding and carries recognised risks that must be weighed in each case. Across the whole subject the consistent themes are that the laxity is generalised but matters most at the top of the neck, that a measurement on a scan and the wellbeing of the spinal cord are only loosely related, that clinical vigilance for the signs of myelopathy has come to occupy the centre of assessment, and that any individual outlook is a matter for the treating team rather than something that can be read from a single number or category.