

Chiari Malformation

A patient guide

In brief: A Chiari I malformation is a structural finding in which the lowest part of the cerebellum — the cerebellar tonsils — sits below the natural opening at the base of the skull. Although it is often described as a brain finding, its greatest significance for a spinal surgeon lies in what it can do further down: it is the commonest cause of a fluid-filled cavity within the spinal cord (syringomyelia), a recognised cause of childhood spinal curvature, and, in a subset of people, it coexists with problems at the junction between the skull and the upper neck. This page approaches Chiari I from that spinal angle.

What Chiari I malformation is (tonsillar descent)

The cerebellum is the part of the brain that sits at the back of the head, tucked into a compartment called the posterior cranial fossa. At the floor of this compartment is the foramen magnum, the large opening through which the brainstem becomes the spinal cord. The cerebellar tonsils are two small, rounded lobes at the bottom of the cerebellum that normally rest just above this opening.

In a Chiari I malformation, the tonsils are displaced downwards (caudally) so that they project through the foramen magnum into the top of the spinal canal. On magnetic resonance imaging (MRI), the convention in adults is that descent of more than approximately five millimetres below a reference line drawn across the foramen magnum (the McRae line) is considered abnormal. A position of up to about three millimetres is regarded as normal, and three to five millimetres as borderline. In children the thresholds are more generous and vary with age, because tonsillar position changes during growth.

Chiari I is distinct from the other forms first catalogued by the pathologist Hans Chiari in 1891. Chiari II is a more extensive malformation, involving the brainstem and the fourth ventricle, and is closely linked with spina bifida (myelomeningocele). A further description, sometimes called Chiari 0, refers to people who have the typical clinical picture and often a syrinx, but little or no measurable tonsillar descent, accompanied by a crowded, small posterior fossa. The unifying theme across these variants — and the reason millimetres alone tell only part of the story — is obstruction to the free movement of cerebrospinal fluid at the base of the skull.

A central point, established in large clinical series, is that the measured position of the tonsils correlates poorly with whether a person has symptoms. Some individuals with marked descent feel entirely well, while others with borderline descent are markedly affected. For this reason, modern understanding has shifted away from a purely millimetre-based definition towards one centred on cerebrospinal fluid dynamics and posterior fossa crowding.

Why it matters to the spine (the link with syringomyelia)

The single most important spinal consequence of a Chiari I malformation is syringomyelia: the formation of a fluid-filled cavity, or syrinx, within the substance of the spinal cord. Chiari I is the commonest cause of this condition. Across published series the proportion of people with Chiari I who also have a syrinx ranges widely, from roughly a quarter to two-thirds, with one of the largest paediatric series reporting a figure of around 57 per cent.

The brain and spinal cord are bathed in cerebrospinal fluid, which normally moves to and fro with each heartbeat as the arterial pulse expands the brain slightly. When the cerebellar tonsils plug the foramen magnum, this pulsatile flow is obstructed. The prevailing modern explanation, developed from intra-operative ultrasound and specialised flow-sensitive MRI, is that the herniated tonsils move downwards in a piston-like fashion with each systole, momentarily sealing the opening. This converts the smooth cerebrospinal fluid pulse into enlarged pressure waves within the spinal subarachnoid space, which compress the cord from the outside and drive fluid

into it, propagating a cavity along the cord with each heartbeat. This account superseded earlier theories that imagined fluid being forced directly down the central canal of the cord (the historical "water-hammer" concept) or sucked in through a one-way valve mechanism.

The practical importance of this model is considerable. Because the syrinx is generated by obstruction at the base of the skull rather than by a problem intrinsic to the cord, the logical operation is one that relieves the obstruction and restores fluid flow at the foramen magnum, not one that simply drains the cavity. It is also why the syrinx, rather than the tonsillar descent itself, is usually the source of symptoms felt in the body — most characteristically a band-like loss of pain and temperature sensation across the shoulders and arms with preserved light touch, together with wasting and weakness of the hands. This pattern reflects damage to the fibres crossing in the centre of the cord and to the anterior horn cells, the structures a centrally placed cavity injures first.

Its association with scoliosis

In children and adolescents, a curvature of the spine is one of the commonest ways a Chiari I malformation with a syrinx first comes to attention. In large paediatric series, scoliosis is a presenting feature in a substantial minority — around 18 per cent in one major series — and in cohorts selected for the presence of a syrinx the proportion is considerably higher.

What distinguishes this kind of scoliosis from ordinary adolescent idiopathic scoliosis is the underlying cause. In adolescent idiopathic scoliosis the curve typically bends towards the right in the thoracic spine and has no demonstrable neurological cause. By contrast, scoliosis driven by a syrinx is more often atypical: a left-convex (left-bending) thoracic curve, an unusually early age of onset, a painful curve, rapid progression, or curves accompanied by neurological signs such as altered abdominal reflexes, sensory changes or hand weakness. Any of these features is a recognised prompt to image the whole neuraxis rather than the spine alone, precisely because a Chiari I malformation and syrinx may be the hidden driver.

The mechanism is thought to be asymmetrical. An expanding syrinx injures the anterior horn cells and sensory pathways unevenly across the cord, producing an imbalance in the paraspinal muscles that pulls the growing spine into a curve. In other words, the deformity is a downstream symptom of a spinal cord lesion rather than a primary problem of the bones and discs.

This has a direct bearing on treatment sequence and is discussed further below: in suitable younger children with smaller curves, relieving the obstruction at the base of the skull can halt or even partially reverse the curvature, whereas a Chiari I malformation without a syrinx has little influence on how a curve behaves. The syrinx, once again, is the operative target.

Its relationship with the craniovertebral junction and instability

The craniovertebral junction is the complex of bone, ligament and joint where the skull meets the first two vertebrae of the neck (the atlas and axis). In most people with a Chiari I malformation the problem is straightforwardly one of a small, overcrowded posterior fossa with downward tonsillar descent, and the craniovertebral junction is stable. In a subset, however, the picture is more complicated, and recognising this group is one of the most consequential judgements a spinal surgeon makes.

Two related abnormalities matter here. The first is basilar invagination, in which the odontoid peg of the second vertebra migrates upwards into the foramen magnum, compressing the front of the brainstem and cervical cord. The second is atlantoaxial instability, in which the joint between the first and second vertebrae is abnormally mobile. These can occur together, and either can coexist with tonsillar descent.

An influential body of work has argued that, in a significant proportion of people labelled as having a Chiari malformation — particularly those with a certain pattern of basilar invagination — the primary disorder is in fact instability at the atlantoaxial joint, and that the tonsillar descent is a secondary or even protective response to it. The therapeutic implication of this view is the opposite of conventional Chiari surgery: rather than removing bone

at the back of the skull to make space, the proposal is to realign and fuse the first and second vertebrae to address the instability directly, on the grounds that removing bone alone might worsen an unstable junction.

This position remains debated, and not every case fits it. The settled, practical message, however, is firm and is shared across the debate: the craniovertebral junction should be deliberately assessed for instability and basilar invagination before a decompression of the back of the skull is assumed to be the right operation. Distinguishing a straightforward small-posterior-fossa Chiari I from disease driven by ventral bony compression and instability is the pivotal decision, because the two demand fundamentally different surgery.

Why it happens

The most widely accepted explanation for Chiari I is a developmental one, centred on the bones of the skull base rather than on the brain itself. The structures that form the floor and walls of the posterior cranial fossa arise from a tissue layer called the mesoderm. If these bones develop slightly under-sized, the resulting compartment is too small to hold a normally formed cerebellum. The hindbrain, having nowhere else to go, is displaced downwards through the only available exit — the foramen magnum. Careful measurements of the posterior fossa in affected people support this account, consistently showing a smaller-than-average compartment with normal-volume contents, hence the description of an "overcrowded" posterior fossa.

This explains why Chiari I is generally present from early life and is frequently discovered by chance, and why it is best understood as a problem of insufficient space rather than of an oversized or diseased brain. Most cases are isolated, although Chiari I can occur alongside certain disorders of bone and connective tissue, and acquired forms can arise when pressure relationships across the foramen magnum are altered by other processes. The common final pathway in all of these is the same: crowding at the foramen magnum and obstruction of cerebrospinal fluid flow.

How it tends to present

The presentation of Chiari I is highly variable, and a large number of people with the finding have no symptoms at all, the malformation being noted incidentally on a scan performed for an unrelated reason.

When symptoms do occur, they fall broadly into two groups. The first relates to the crowding at the base of the skull itself. The most characteristic feature is a headache felt at the back of the head and upper neck that is brought on or worsened by coughing, sneezing, laughing or straining — manoeuvres that briefly raise pressure and accentuate the obstruction. Neck pain is also common. Some people describe dizziness, unsteadiness, difficulty swallowing, hoarseness or disturbances of balance and eye movement, reflecting pressure on the brainstem and lower cranial nerves.

The second group of symptoms relates to a syrinx within the spinal cord, and these are the features of greatest interest from a spinal perspective. They include a dissociated loss of sensation — reduced perception of pain and temperature across the shoulders and arms while light touch is preserved — together with wasting, weakness and clumsiness of the hands, and sometimes pain. In children, as described above, the first sign may instead be a developing curvature of the spine, with the sensory and motor features either subtle or not yet apparent.

The breadth of this picture, and the fact that some of the most important features are spinal rather than cranial, is the reason imaging of the whole spine, and not only the head, is central to assessment.

How it is assessed (MRI, cine CSF flow)

Magnetic resonance imaging is the cornerstone of assessment. Sagittal images of the brain and craniocervical junction demonstrate the position and shape of the cerebellar tonsils relative to the foramen magnum and allow the degree of descent to be measured. Crucially, imaging is not confined to the head: MRI of the whole spine is regarded as essential, both to detect and characterise a syrinx and to investigate any spinal curve, since the cavity may extend over many segments and may be the true source of symptoms.

A specialised sequence known as phase-contrast cine, or cerebrospinal fluid flow, imaging adds a dynamic dimension. Rather than showing anatomy alone, it captures the to-and-fro movement of fluid at the foramen magnum across the cardiac cycle and can demonstrate where flow is obstructed. Abnormal or reduced flow supports the view that a malformation is clinically relevant — particularly valuable where the tonsillar descent is borderline, or in the Chiari 0 pattern where descent is minimal — and improvement in flow after surgery has been observed to track with improvement in symptoms. It is regarded as a useful adjunct that informs judgement rather than a stand-alone diagnostic test.

Where instability or basilar invagination is suspected, assessment of the craniovertebral junction is extended. This may include computed tomography to define the bony anatomy in detail and imaging in flexion and extension to look for abnormal movement between the vertebrae. As set out above, this step is what allows the unstable, ventrally compressed group to be separated from straightforward Chiari I before any decision about surgery is reached.

How it is distinguished from related conditions

Three distinctions are particularly important, and confusion between them has direct consequences.

The first is between Chiari I and syringomyelia. These are not the same thing, although they frequently occur together. Chiari I is an anatomical cause — tonsillar descent obstructing fluid flow at the foramen magnum. Syringomyelia is one of its consequences — a fluid cavity within the cord. Each can exist without the other: a syrinx may arise from quite different causes, such as previous injury to the cord, scarring of the lining membranes, or a tethered cord, and a Chiari I malformation may be present with no syrinx at all. Establishing which is present, and whether the Chiari is responsible for the syrinx, shapes the entire approach.

The second is between Chiari I and basilar invagination. In Chiari I the essential problem is a small posterior fossa with tonsillar descent from above. In basilar invagination the problem is bony: the odontoid peg rides upwards into the foramen magnum, compressing the brainstem and cord from the front, and is often accompanied by instability of the upper neck. The directions of compression differ, the role of instability differs, and — as discussed — the operations differ, sometimes diametrically. This is why the two are deliberately separated during assessment rather than grouped together as a single "Chiari" problem.

The third is between Chiari I and Chiari II. Chiari II is a more extensive hindbrain malformation associated with spina bifida and is a different clinical entity with its own management, distinct from the isolated tonsillar descent of Chiari I that is the subject of this page.

What the evidence shows about management (observation vs posterior fossa decompression) — impersonal

Where a Chiari I malformation causes no symptoms, or only mild ones, the weight of evidence indicates a generally benign course. Studies following people managed without surgery — including some with a stable syrinx — report that the large majority remain stable over time. In this setting, monitoring with periodic MRI is a recognised and widely adopted approach, with surgery reserved for progressive symptoms, an enlarging or newly symptomatic syrinx, or a progressing spinal curve.

When surgery is indicated, the established operation is posterior fossa decompression, also called foramen magnum decompression. Its aim is to enlarge the cramped compartment at the back of the skull and restore the free movement of cerebrospinal fluid across the foramen magnum, thereby addressing the mechanism that generates a syrinx. A persistent technical question in the literature concerns whether the dura — the tough membrane covering the brain and cord — should be opened and enlarged with a patch (a procedure termed duraplasty). The evidence, drawn from several systematic reviews and meta-analyses, suggests a trade-off: decompression with duraplasty tends to give better improvement in symptoms and in the syrinx, and a lower rate of repeat surgery, but carries a higher rate of fluid-related complications such as leakage and inflammation of the

lining membranes. Decompression without opening the dura is quicker and lower in morbidity, with comparable results in people who do not have a syrinx. A pragmatic synthesis frequently described in the literature is that the presence of a syrinx favours opening and enlarging the dura, whereas its absence often makes a bony decompression alone sufficient. Intra-operative ultrasound is increasingly used to judge the adequacy of decompression as it is performed.

Two further situations call for different operations. Where a syrinx persists despite measures aimed at restoring fluid flow, or where syringomyelia arises from scarring of the lining membranes rather than from Chiari I, surgery directed at the cavity itself, or microsurgical clearance of the scar tissue, may be considered; the presence of such scarring is a recognised predictor of recurrence. And where instability or basilar invagination is the true driver, the literature describes stabilisation and fusion of the upper neck rather than, or in addition to, decompression. In all of these, the choice depends on the individual configuration of the problem and is determined by the treating specialist team.

Outlook

For the majority of people, particularly those who are without symptoms or have only mild ones, the outlook associated with a Chiari I malformation is reassuring, and the finding remains stable under observation. Where a syrinx and symptoms are present and surgery is undertaken to restore fluid flow, many people experience stabilisation or improvement of their symptoms, and the syrinx often reduces over the months that follow. In children, scoliosis driven by a syrinx may halt or partly reverse after timely decompression when the curve is still small and the child is young, although larger or more established curves frequently require separate deformity treatment in addition. Outcomes vary with the underlying configuration — the size and extent of any syrinx, the presence or absence of instability or basilar invagination, and the age at which a curve is addressed — which is why careful characterisation of each of these features is central to anticipating the course.