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SPECIAL ANNUAL ISSUE

# Chiari I—a 'not so' congenital malformation?

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**Abstract**
The term Chiari I malformation (CIM) is imbedded in the paediatric neurosurgical lexicon; however, the diagnostic criteria for this entity are imprecise, its pathophysiology variable, and the treatment options diverse. Until recently, CIM has been considered to be a discrete congenital malformation requiring a uniform approach to treatment. Increasingly, it is recognised that this is an oversimplification and that a more critical, etiologically based approach to the evaluation of children with this diagnosis is essential, not only to select those children who might be suitable for surgical treatment (and, of course those who might be better served by conservative management) but also to determine the most appropriate surgical strategy. Whilst good outcomes can be anticipated in the majority of children with CIM following foramen magnum decompression, treatment failures and complication rates are not insignificant. Arguably, poor or suboptimal outcomes following treatment for CIM reflect, not only a failure of surgical technique, but incorrect patient selection and failure to acknowledge the diverse pathophysiology underlying the phenomenon of CIM. The investigation of the child with 'hindbrain herniation' should be aimed at better understanding the mechanisms underlying the herniation so that these may be addressed by an appropriate choice of treatment.

**Keywords** Chiari I · Congenital malformation · CNS manifestations · Cerebellar tonsillar ectopia · Hindbrain hernia · Craniovertebral junction

## Introduction

There are few conditions in the entire practice of paediatric neurosurgery that are shrouded in such controversy and that evoke such diversity of opinion than is the case for Chiari I malformation (CIM). The controversies encompass terminology, definition, pathophysiology and indications for treatment as well as surgical technique. In the majority of cases the 'entity' that we now call 'Chiari I malformation' bares little relationship to the condition as described by Hans Chiari, indeed evidence continues to accrue that seems to contradict the notion that this is a congenital malformation at all. These are more than semantic nuances; failure to recognise the etiological heterogeneity that underlies CIM may result in incorrect treatment selection and compromise outcomes.

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## Terminology—'malformation versus deformation'

"On changes in the cerebellum, pons and medulla oblongata due to congenital hydrocephalus of the cerebrum" (Fig. 1). In his 1892 monograph, Hans Chiari described the three malformations that bare his name; he added type IV subsequently [1]. As indicated by the title, Chiari considered that these malformations were not only congenital, but that hydrocephalus was an important etiological factor. Type III and type IV Chiari malformations are rare in clinical practice; both comprise significant malformations of the hindbrain (typically in the context of occipital or occipito-cervical meningoencephalocele) and hydrocephalus is indeed a frequent association in these types. Type II Chiari malformation is of course more common than types III and IV and is characteristically seen in the context of myelomeningocele, where there is also a strong association with hydrocephalus. Chiari types II, III and IV can each be considered veritable, congenital malformations, with pan CNS manifestations. In Chiari II for example, in addition to the extensive changes in the rhombencephalic derivatives, there are widespread supratentorial changes including fusion of the mass intermedia, interdigitation of cortical gyri and 'beaking' of the tectum.


ÜBER

VERÄNDERUNGEN DES KLEINHIRNS, DES PONS UND DER MEDULLA OBLONGATA

IN FOLGE VON

CONGENITALER HYDROCEPHALIE DES GROSSHIRNS

VON

Dr. H. CHIARI,

WIEN 1895.

**Fig. 1** Title page from Hans Chiari monograph [1]

By contrast, what Chiari described as CIM is typically a focal abnormality, comprising prolapse of the cerebellar tonsils, sometimes with distortion of the brainstem at the level of the foramen magnum, usually associated with chronic hydrocephalus but lacking the more widespread brain changes that characterise the other types. It is both unfortunate and inaccurate that the entity of CIM, as described by Chiari (in autopsy studies!), has been extrapolated to encompass almost all forms of cerebellar tonsillar ectopia encountered in contemporary paediatric neurosurgical clinical practice.

Whilst Chiari types II, III and IV are indeed true congenital malformations of the central nervous system, the same cannot be said of CIM. Some authors have used the terms 'hindbrain hernia' or 'cerebellar tonsillar ectopia' in an attempt to emphasise that CIM should be more appropriately considered an acquired 'deformation' of the normal hindbrain rather than a true 'malformation' (Fig. 2). There are a number of lines of argument that can be cited to support this assertion. Firstly, CIM is rarely seen in the newborn; when 'cerebellar tonsillar ectopia' does appear in early childhood, this is most likely due to a discrepancy between postnatal growth of the cerebellum and that of the posterior cranial fossa. Secondly, histological analysis of cerebellar tonsils in Chiari I is either normal or shows areas of pressure necrosis [2] rather than the neuronal disorganisation and brainstem dysplasia that have been described for example in Chiari II malformation. Thirdly, there are numerous examples in the literature that demonstrate the reversibility of cerebellar tonsillar ectopia following treatment of the underlying cause and that the morphology of the hindbrain and tonsil will often normalize following foramen magnum decompression [3].

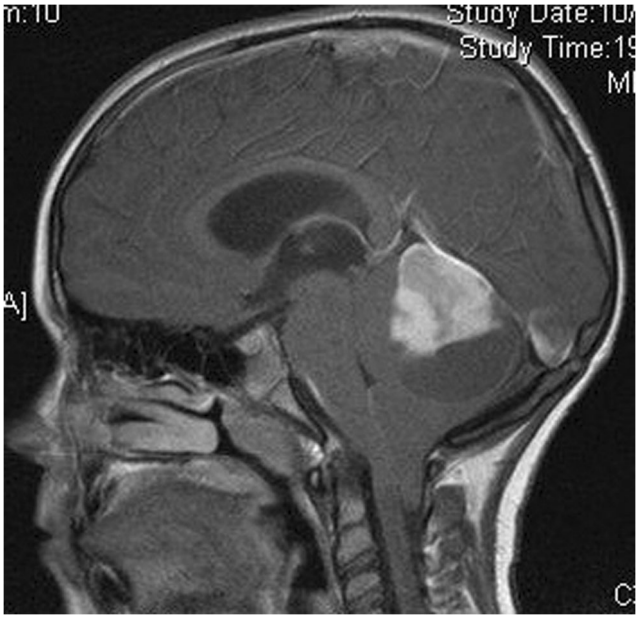

**Fig. 2** T1 weighted, post contrast sagittal MRI scan showing cerebellar tonsillar ectopia caused by 'deformation' of the cerebellum due to an intrinsic tumour

Whilst the term CIM will continue to be used in this chapter, the reader should perhaps consider that CIM is generally an acquired, and potentially reversible anomaly, whilst the other Chiari malformations are congenital and irreversible.

## Definition

There is no agreed definition of CIM. The most commonly cited definition is decent of the cerebellar tonsils of at least 5 mm below the foramen magnum, although definitions range from 0 mm (tonsils at the level of the foramen magnum) to 1.5 cm. Defining CIM by measurement is not only arbitrary but potentially misleading, risking both over-diagnosis and under-diagnosis. The reasons for this are as follows: the plane of the foramen magnum is not flat but ellipsoid or 'saddle shaped', thus making it difficult to establish an appropriate point of reference for the measurements. Additionally, since the tonsils are para median structures, yet measurements are usually taken from a midline sagittal MRI scan, this measure will not necessarily reflect the true extent tonsillar decent [4, 5]. A further complicating factor is the variability in the level of the cerebellar tonsil found in the normal population [6]. Barkovitch found that in 14% of normals, the tonsils were at or below the level of the foramen magnum [7]. Finally, there is no clear correlation between the extent of tonsillar herniation and the likelihood of symptoms or syrinx formation; indeed, the entity of Chairi 0 was introduced to account for those patients with syrinx but with cerebellar tonsils at or above the foramen Magnum whose syringes improved after a foramen magnum decompression operation [8, 9].

The introduction of terms such as Chiari 0 and Chiari 1.5 [10] whilst referring to recognisable radiological entities is apt to further confuse rather than clarify the situation. These additional "grades" of Chiari malformation tend to propagate the incorrect notion of a spectrum of anomaly, ranging from mild CIM to severe (types II, III and IV).

In summary, including a numerical measure in the definition of CIM is not only imprecise but it lacks clinical relevance and should probably be abandoned. A more useful definition of CIM might be that of 'a functional obstruction at the foramen magnum by the inferior cerebellum; one that may result in neuraxis compression and/or obstruction to CSF flow across the craniovertebral junction'. Such a definition not only obviates the need for a measurement, but allows for the possibility of other pathophysiological mechanisms that might result in compromise of this region, for example local scarring of the subarachnoid space or bony malformation at the craniovertebral junction. Thus, Chiari I, Chiari 0 and Chiari 1.5 would all be included under such as definition, each representing some degree of acquired obstruction at the level of the foramen magnum.

## Pathophysiology of Chiari I malformation

Instead of considering the CIM as a single diagnostic entity, herniation of the hindbrain should be viewed as the consequence of different pathophysiological processes. Of course a clear, underlying predisposition might not be possible to identify in all cases, leaving an entity of 'idiopathic CIM' still best treated by foramen magnum decompression. However, an attempt to refine more precisely the cause of the herniation opens the possibility for an algorithmic approach to investigation and appropriately tailored treatment.

## Chiari I—a disorder of CSF circulation

In Chiari's original monograph, with reference to the C1M, he makes the following statement "I have had the impression that the extension of the tonsils and medial sides of the inferior lobes probably always is the result of chronic and very early onset of cerebral hydrocephalus. Therefore the term congenital hydrocephalus must be accepted. I have found it in a relatively large parentage of cases of chronic congenital hydrocephalus, but never without hydrocephalus or in cases of acute hydrocephalus or later developing hydrocephalus" [1]. Two factors need to be bourne in mind when considering Chiari's first type of malformation. Firstly, his observations were based on post mortem specimens; his examples likely representing advanced stages of disease, far removed from the scenario seen in paediatric neurosurgical practice today. Secondly, the association with hydrocephalus in a 'large percentage of cases' is at variance with the observations from contemporary clinical and radiological literature; overt hydrocephalus with

ventriculomegaly occuring in only 10% of paediatric patients with C1M [11].

Most neurosurgeons concur that when overt hydrocephalus occurs in the presence of C1M, the hydrocephalus should be treated first [12]. Improvement in tonsillar herniation, or indeed complete reversal of the C1M following hydrocephalus treatment, whether by shunt or endoscopic third ventriculoscopy, is now well documented (Fig. 3). Massimi et al. reported improvement in both hydrocephalus and C1M symptoms in all of 15 patients who had been treated with ETV. Mean tonsillar herniation was 12.7 mm (range 6–35 mm) preoperatively and 8.3 mm (range 3–27 mm) post-operatively [13]. None of the patients required posterior fossa decompression. In a similar study, 16 patients with C1M and hydrocephalus were treated with ETV, of those with C1M symptoms half were improved and syringomyelia resolved in 60%. Six patients in this series ultimately required posterior fossa decompression for persisting C1M symptoms [14].

These two series concern CIM patients where there is ventriculomegaly and symptoms of raised ICP, where the indication to address hydrocephalus primarily is straightforward. However, more subtle disorders of CSF circulation may accompany CIM and might also benefit from ICP directed therapy. Tonsillar ectopia is reported in up to 20% of cases of idiopathic intracranial hypertension. In a retrospective study of 192 patients who had undergone posterior fossa decompression for CIM, 36 did not improve and of these 41.6% were found to have a pseudo tumour syndrome that subsequently responded well to CSF diversion [15]. The potential importance of impaired brain compliance in the aetiology of CIM was raised in a recent study of adult patients evaluated using ICP monitoring. Continuous ICP measurements were made in 16 patients with symptomatic C1M, whilst all had mean ICP values in the normal range, 14/16 showed increased ICP pulsatility suggesting impaired intracranial compliance. Six of these patients were treated with primary ventriculoperitoneal shunt insertion with symptomatic improvement [16].

Although the CIM seen in the majority of paediatric patients seems to bare little resemblance to the condition as described by Chiari, the relationship between hydrocephalus (or, perhaps more correctly abnormal CSF circulation) and CIM might be underestimated. Whilst overt hydrocephalus occurs in a relative minority of CIM patients, disordered CSF circulation may be more prevalent than previously believed. Indeed failure to acknowledge the role of abnormal CSF circulation might account for treatment-related morbidity, including some cases of post-operative CSF leak, pseudomeningocele and hydrocephalus, following CIM surgery. A more proactive approach to evaluating ICP as part of the early evaluation of children with symptomatic CIM should perhaps be considered.

## Chiari I—a disorder of the craniovertebral junction

One of the prevailing theories for the pathogenesis of C1M has been that of craniocerebral disproportion; caudal displacement of the hindbrain structures due to reduced posterior fossa volume. However, the evidence to support this assertion has been contradictory; some studies confirming reduced posterior fossa volume in cases of C1M [17, 18] whilst others failing to demonstrate a consistent relationship [19, 20]. This is perhaps unsurprising, particularly in the paediatric population in whom the growth rates of the neuraxis and surrounding bone structures are neither linear over time nor parallel. Furthermore, many early estimates of posterior fossa volume were extrapolated from linear measurements that, in the light of contemporary automated techniques, have been shown to correlate poorly with posterior fossa volume [21].

More recently, attention has focused on abnormal biomechanics and anatomy of the craniovertebral junction (CVJ) rather than posterior fossa volume. It has long been recognised that certain bony malformations such as atlanto-occipital assimilation, basilar invagination and craniocervical instability occur more commonly in cases of CIM yet their potential significance has hitherto been underestimated and has not been incorporated into surgical decision-making; sometimes

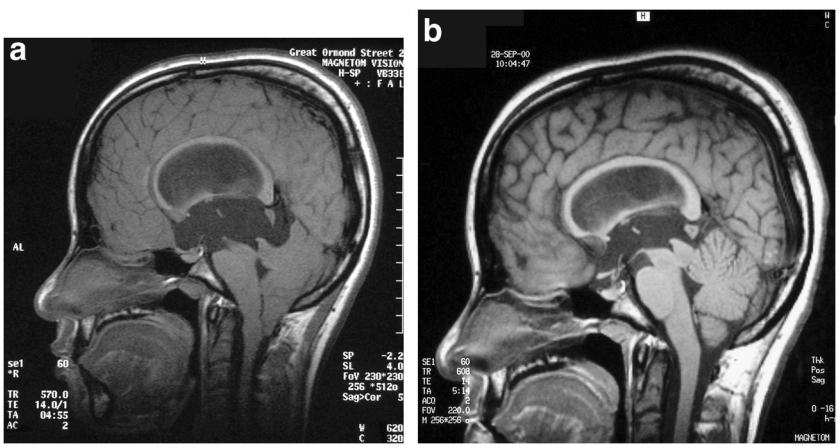


**Fig. 3** Sagittal MRI scans illustrating Chiari I malformation due to hydrocephalus (**a**). Reversal of Chiari I malformation following endoscopic third ventriculostomy (**b**)

with deleterious consequences (Fig. 4). Among 45 C1M patients who required revisions of foramen magnum surgery due to sub optimal outcomes after primary surgery, Klekamp identified 10 patients with craniocervical instability [22].

The combination of descent of the obex in addition to tonsillar ectopia referred to as the Chiari1.5 malformation seems to indicate a less predictable response to treatment. In one series, re-operation for persisting syringomyelia, following initial conventional foramen magnum decompression was required in 13.6% of Chiari 1.5 cases. Bony abnormalities at the CVJ are more commonly seen in cases of Chiari 1.5 compared with 'standard' CIM; these include basilar invagination, atlas assimilation and varying degrees of odontoid retroflexion. It is now recognised that these patients are among the subset of CIM patients who benefit from concomitant decompression and craniocervical fixation.

How common is the need for craniovertebral fusion in C1M? In recent large series of C1M, between 10 and 20% of cases required fixation procedures in addition to decompression [23, 24]; however, surgical series are likely to be biased by specific institutional philosophy. In another series of 500 cases, only 4 (1%) required fusion. Can the need for concomitant decompression and fixation be predicted? In a detailed retrospective review of paediatric cases that had undergone surgery for C1M, Bollo et al. identified radiological parameters that increased the likelihood of occipito-cervical fixation being required. These factors were descent of the obex as well as the tonsils, below the plane of the foramen magnum (Chiari 1.5), basilar invagination, and a clivo-axial angle < 125 degrees [23]. For this group of patients deemed at high risk for failure following conventional decompression, occipito-cervical fixation should be considered as a primary treatment. They used the term 'complex Chiari' for this subgroup of C1M patients (Fig. 5).

In a more controversial publication, Goel proposed that atlantoaxial instability was the primary cause of CIM, whether or not associated with basilar invagination. The criteria for instability in his study were based on a novel classification of anomalous facet alignment, rather than conventional measures of instability [25]. Goel went on to advocate distraction of the C1/C2 facet joint with atlantoaxial arthrodesis as the treatment of choice, additionally suggesting that neither foramen magnum decompression nor incorporation of the occiput into the fusion construct was necessary. Close inspection of

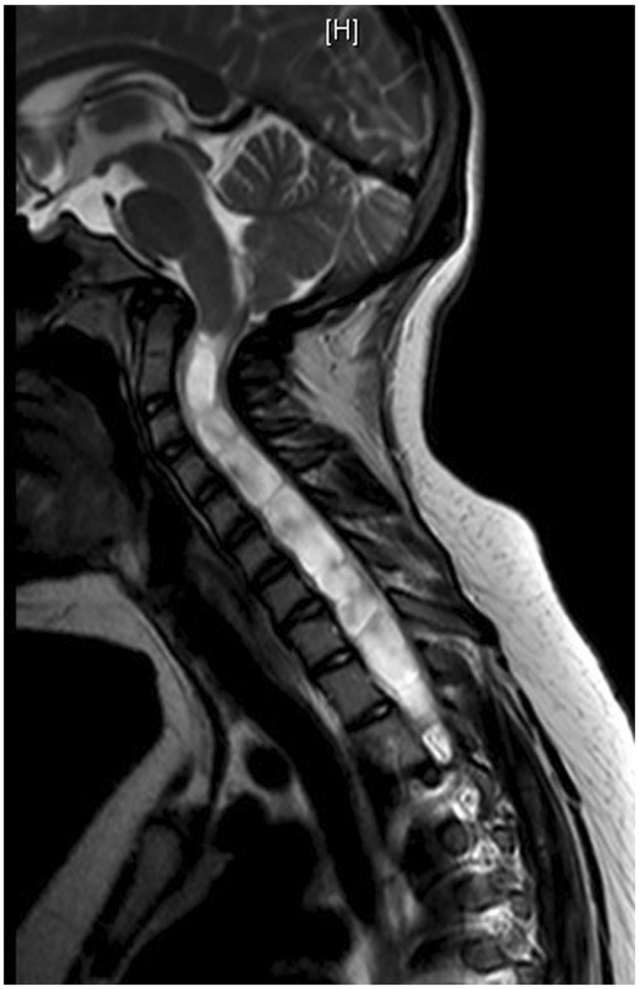


**Fig. 4** Syringomyelia due obstruction at the foramen magnum secondary to short clivus and platybasia (sagittal T2-weighted MRI scan)

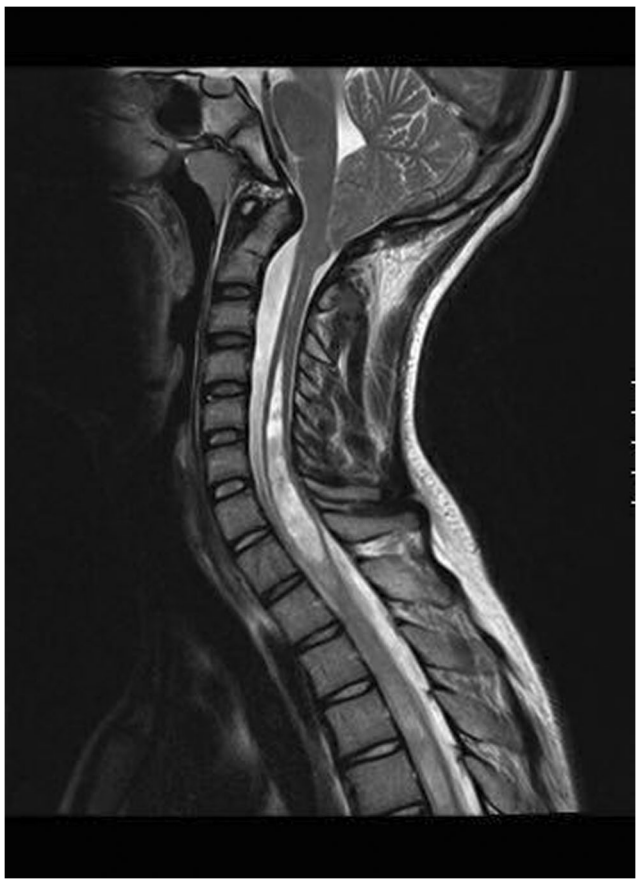

**Fig. 5** Complex Chiari malformation. There is Chiari 1.5 malformation (descent of the obex and cerebellar tonsils) with basilar invagination and acute clivo-axial angle (sagittal T2-weighted MRI scan)

his patient cohort suggests that many of the patients in that study harboured significant CVJ malformations, and that the results might not be applicable to the wider Chiari population. Excluding the occiput from fixation constructs, particularly when distraction has been necessary to redress the craniovertebral malalignment, is at variance with prevailing practice.

Is there a role for ventral decompression? Clival abnormalities and retroflexion of the odontoid are common in CIM and may distort and compress the brainstem, indeed the incidence of odontoid retroflexion in paediatric CIM is approximately 80% [26, 27]. Therefore, the potential need to address ventral as well as posterior compression at the CVJ in cases of CIM not uncommonly arises. The algorithm originally proposed by Menezes was based on whether the CVJ abnormality was reducible or irreducible; irreducible cases being selected for trans-oral decompression as part of the surgical strategy. In an early series of adult and paediatric CIM patients with basilar invagination, preliminary trans-oral decompression was performed in 66/100 cases [28]. Ventral decompression is not without risk and does not obviate the need for a posterior procedure (fixation with or without decompression). The degree of odontoid retroflexion and its effect on the neuraxis are most commonly expressed by the Grabb measurement or pB-C2 line [29]. In his original publication, Grabb found that ventral decompression was not required if this measure was < 9 mm. Increasingly, it is recognised that traction (prior to or during surgery) will adequately address ventral compression in the majority of cases, obviating the need for trans-oral surgery [30].

## Chiari I—a disorder of intracranial venous drainage and craniosynostosis

The interaction between venous hypertension, intracranial pressure and cerebrospinal fluid circulation is complex; disturbances in each of these have been correlated with CIM.

One of the earliest demonstrations of the association between CIM and intracranial venous hypertension came from observations in vein of Galen malformation where it was suggested that engorgement of the brain parenchyma due to raised venous pressure, resulted in extrusion of the cerebellar tonsils through the foramen magnum (Fig. 6). In support of this hypothesis, the CIM was shown to reverse following successful treatment of the vascular anomaly [31]. It is noteworthy that reversibility of the CIM was only observed when adequate venous drainage from the posterior fossa was established after therapeutic embolisation.

Evidence from other sources suggests that anomalous posterior fossa venous drainage might be applicable to a wider group of CIM patients. A recent pilot study of adult CIM patients sought to identify specific MRI parameters that were predictive of clinical outcome following CIM surgery. Reduced jugular venous drainage was among the strongest predictors of poor outcome [32].

In syndromic craniosynostosis, the potential role of abnormal posterior fossa venous drainage in the aetiology of CIM has received much attention (Fig. 7). CIM is present in almost all cases of Kleeblatschadel (cloverleaf skull), it is also extremely common in Crouzon syndrome (70%) and Pfeiffer syndrome (50%) but distinctly unusual in Apert syndrome (< 2%). Cinalli et al. suggested that these differences could be explained on the basis of the pattern of closure of the skull base sutures, in particular the lambdoid sutures. In Crouzons syndrome, these sutures close early in infancy, at a time of rapid postnatal growth of the posterior fossa contents, resulting in a state of craniocerebral disproportion and secondary hindbrain herniation. By contrast in Aperts syndrome, the sutures were seen to persist for longer permitting adequate growth to accommodate the maturing cerebellum [33].

This hypothesis whilst attractive is likely to be an oversimplification and subsequent studies have failed to show a consistent correlation between posterior fossa volume and CIM in syndromic craniosynostosis, indicating that other mechanisms need to be explored [34]. Disordered posterior fossa venous

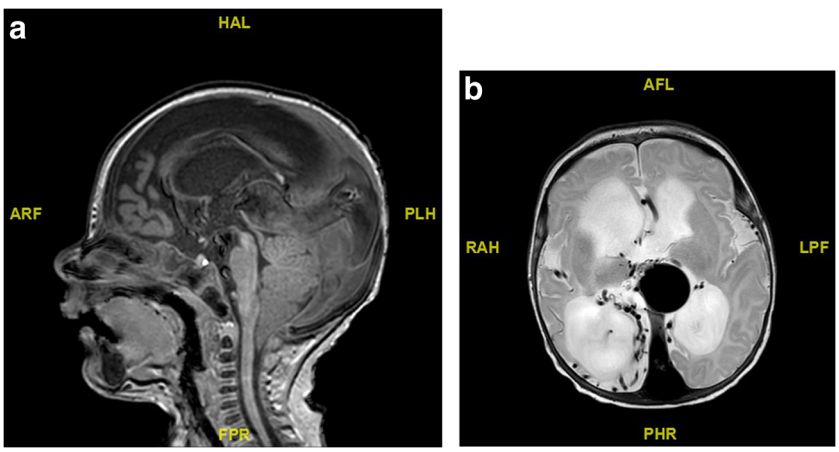


**Fig. 6** **a** Sagittal T1-weighted MRI scan demonstrating Chiari I malformation due to Vein of Galen malformation. **b** Axial T2-weighted MRI showing vein of Galen malformation

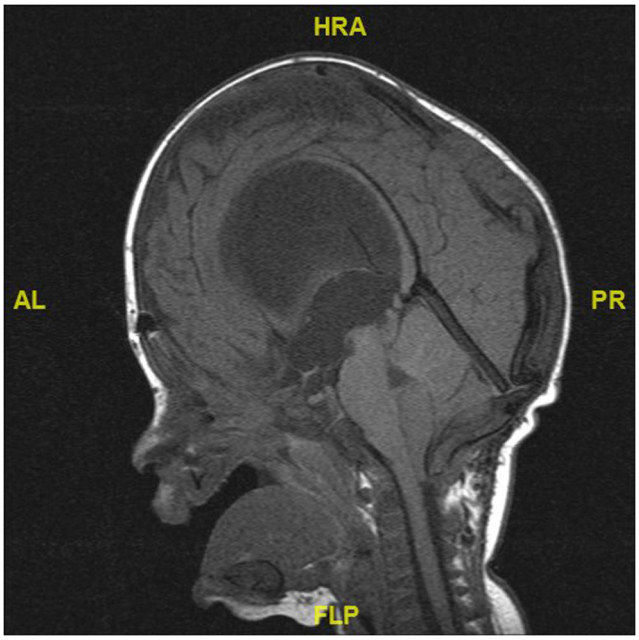


**Fig. 7** **a** Chiari I malformation in a child with Pfeiffer syndrome. There is a small posterior fossa, ventricular enlargement and abnormal skull shape (sagittal T1-weighted MRI scan). **b** CT venogram in the same patient showing prominent paraspinal venous plexus

drainage is a promising candidate. Jugular foraminal stenosis and collateral venous drainage are much more common among cases of syndromic craniosynostosis compared with controls [35, 36]. In one angiographic study, 18/23 patients with craniosynostosis had either complete or more than 50% occlusion of one or both sigmoid/jugular sinuses. The intracranial venous hypertension induced by these changes (frequently exacerbated by chronic $CO_2$ retention due to upper airways obstruction) is thought to propagate a state of increased brain "turgor" resulting in progressive extrusion of the hindbrain though the foramen magnum; a mechanism not dissimilar to that in vein of Galen malformation. Collateral venous drainage may be so florid that it may preclude safe surgical access to the posterior fossa [37].

Increasingly reports are appearing that demonstrate improvement in hindbrain herniation (including syringomyelia) following cranial vault expansion [38, 39]. This surgery, used to address elevated intracranial pressure in the early management of craniosynostosis, leads to improved intracranial compliance. The surgery precludes the need for suboccipital decompression, permits preservation of important collateral venous drainage and reduces the risk of major blood loss. Many craniofacial centres now advocate the use of cranial vault expansion as the initial treatment for CIM in complex craniosynostosis.

## Chiari I—a disorder of growth and metabolism

Various disorders of skeletal growth and metabolism have been described in the context of cerebellar tonsillar extopia and have been postulated to have a causative role. This is in keeping with the notion that the Chiari I malformation is commonly a disorder of mesechyme rather than neural tissue; more a disorder of the 'container than the contents'.

### Growth hormone deficiency

Numerous reports have highlighted an association between growth hormone deficiency (GHD) and CIM. In one MRI study of patients with GHD, 20% of cases were found to have CIM [40]. The initial supposition was that GHD led to reduced skull growth and thus smaller posterior fossa volume. However, in a detailed morphometric analysis of the posterior fossa in GHD patients, Tubbs et al. found no difference in posterior fossa volume between cases of GHD (with or without CIM) and controls [41]. This study did, however, find that the basiocciput tended to be shorter, and the foramen magnum longer in cases of GHD, factors that might predispose to CIM. The causal association between GHD and CIM is further strengthened by observations of improvement, not only in CIM but also syringomyelia, when GHD patients are treated with growth hormone [42]. There are however many instances where CIM appears to have been precipitated by GH treatment; therefore, the association is far from clear cut.

### Calcium and phosphate metabolism

Rickets is characterised by impaired bone growth and quality and may affect cranial growth. Whilst most commonly due to vitamin D deficiency, Rickets can occur due to excessive urinary loss of phosphate, so-called hypophosphataemic rickets. CIM with or without syringomyelia is well described in these cases (Fig. 8). CIM was identified in 44% of cases of familial hypophosphataemia in one series [43]. A more recent study of children with X-linked hypophosphataemic found an incidence of 25% of CIM [44]. This latter study also revealed a high incidence (55%) of secondary craniosynostosis and so the factors predisposing to cerebellar tonsillar ectopia are likely complex these cases. These findings have led to the suggestion that screening for hypophosphataemia and vitamin D deficiency should become part of the initial evaluation of children with supposedly idiopathic CIM. It remains to be seen whether medical management of the underlying disease can modify the natural history of the CIM.

### Radiation

In an interesting perspective on impaired skull base growth and its association with CIM, Aquilina et al. reported on 10 children with CIM who had received radiotherapy for sellar region tumours [45]. They observed that the annual growth rate of the clivus was reduced after radiotherapy and this predisposed toward the development of CIM. This study and

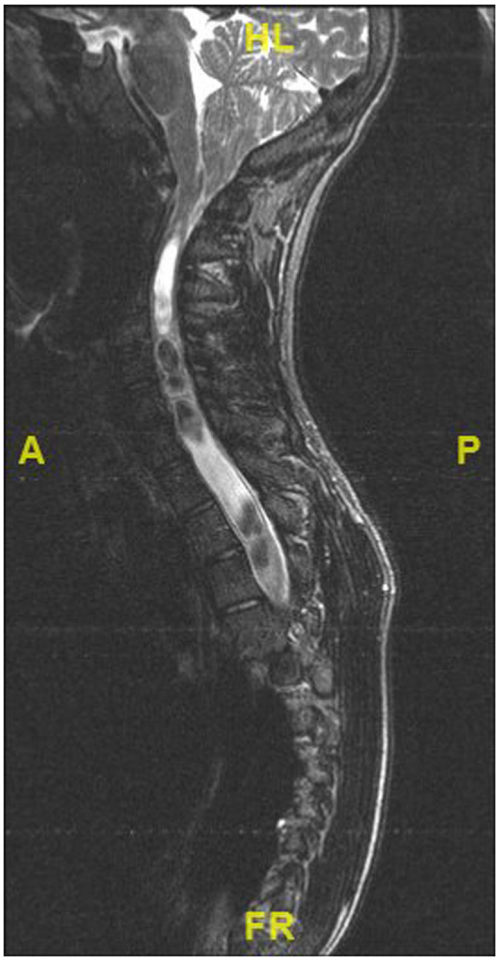


**Fig. 8** T2-weighted MRI scan illustrating Chiari I malformation with syrinx in a 14-year-old boy with hypophosphataemic rickets

previous case reports documenting the same association suggested that young children (under 5 years) are particularly at risk.

### Metabolic bone diseases

A 'mesenchymal' aetiology for CIM is further strengthened by the observation of CIM occurring in a variety metabolic bone diseases including Pagets disease, fibrous dysplasia, Gorehams disease and osteopetrosis. Although it is often unclear whether the effect is mediated simply through reduced growth, or whether factors such as venous hypertension and secondary craniosynostosis, which commonly coexist these conditions, may also have a role.

### Chiari I—a disorder of controversial aetiology

The past 15 years have seen the emergence of a number of controversial associations with CIM. These associations include the occult tethered cord syndrome (OTCS), hypermobility states, in particular hereditary disorders of connective tissue (notably Ehlers-Danlos syndrome, EDS) and the postural orthostatic tachycardia syndrome (POTS). An, arguably concerning, trend in many of these situations has been the tendency to broaden diagnostic criteria (for both tethered cord and CIM) and thus widen indications for surgery. Furthermore, this has led to the emergence of controversial surgical strategies for CIM. The following examples serve to exemplify this.

### CIM and occult tethered cord syndrome

The occult tethered cord syndrome (OTCS) refers to a scenario in which there are clinical features compatible with a tethered spinal cord (sphincter dysfunction, low back pain, lower limb symptoms) yet on MRI, the conus is in a normal position and there are no overt features of spinal dysraphism. Extrapolating from the 'traction hypothesis' for Chiari II malformation, some authors have sought to explain certain instances of CIM in terms of traction on the terminal spinal cord by the film terminale [46, 47]. These authors report favourable outcomes, in both adult and paediatric cases, following simple section of the film; in 9/20 cases described by Royo-Salvador, section of the film was carried out without opening the dura.

Whether the OTCS exists at all, and furthermore, whether there is any association between this and CIM is far from clear. A small, randomised controlled pilot study looking at urological outcomes in cases of OTCS found no objective difference between medical management with or without section of the filum [48].

In the large surgical series of Milhorat, symptom resolution or improvement was reported in over 80% of cases (93% in paediatric cases). In addition, they reported ascent of both the conus and cerebellar tonsils on post-operative MRI. The study concluded that "Simple SFT [section of the film terminale] may be effective in relieving symptomatology, restoring brainstem length, normalising the position of the cerebellar tonsils, and, in many cases, avoiding the need for posterior fossa surgery" This study however needs to be interpreted with caution; the patient population was heterogeneous, and the follow up relatively short (6–27 months). In addition to cases of CIM, defined as cerebellar descent of > 5 mm, they widened their diagnostic criteria to include a group of patients with low lying cerebellar tonsils (0–4 mm). The radiological measurements in this study were made by a single observer and it does not appear that the marginal post-operative measurements (mean = 5 mm ascent of the conus and mean = 3.8 mm ascent of the cerebellar tonsils) were tested for reproducibility.

Whether it is possible for the filum (usually less than 2 mm diameter), in the context of a normally positioned conus, to exert tension sufficient to cause traction of the brainstem is questionable. The spinal cord is, after all, not simply anchored by the filum alone but is attached to the spinal canal along its

length via the nerve roots and dentate ligaments; therefore, any (likely small) tractional forces emanating from the filum will be exerted over a limited number of segments of the lower spinal cord. Furthermore, in situations where the spinal cord is unquestionably low lying (for example lumbosacral lipomas), the hindbrain and cerebellum are typically normally positioned. In a review of the available literature, Massimi et al. [49] found no convincing evidence to support the association between CIM and OTCS.

## CIM and hypermobility

In 2007, Milhorat and colleagues reported an association between hereditary disorders of connective tissue and CIM. In that series, 35% of patients with this combination had a history of failed Chiari surgery [50]. The authors proposed that hitherto unrecognised occipito-atlantoaxial hypermobility was present in this population and that this warranted craniocervical fixation.

Over the past decade, a number of countries have seen increasing numbers of patients diagnosed with hypermobility disorders; these patients frequently have diverse and complex clinical symptoms that are often refractory to medical therapies. Other clinical entities such as the postural orthostatic tachycardia syndrome and irritable bowel syndrome have also become incorporated into this symptom complex as an underlying dysautonomia due to brainstem deformation has been proposed.

Unfortunately, many of the diagnostic criteria for craniovertebral instability in this patient group, frequently necessitating dynamic or upright MRI studies, are not validated and the evidence to support a causal relationship between hypermobility and CIM is currently incomplete. A recent evidence-based analysis found no evidence to support the use of upright MRI over conventional MRI in the evaluation of patients with EDS and suspected craniovertebral pathology [51]. Whilst studies using upright MRI comparing patients with controls are reported to show differences in various craniovertebral morphometric parameters, the differences are often marginal and likely lack the sensitivity to discriminate between hypermobility that lies at the upper end of normal, from true pathological instability that might threaten neurological function [52].

Ehlers-Danlos (hypermobility type) is the most common underlying diagnosis among these patients. Although a causative genetic mutation has been established for the other types of EDS this is not so for the hypermobility type, where diagnostic criteria are essentially clinical. The Beighton score is a composite score obtained by assessing range of movement across various joint and is an important component of the diagnosis. Unfortunately, the specificity of this score is poor. In one recent study of normal adolescents, generalised joint hypermobility was present in 60.6% of girls and 36.7% of boys using a Beighton score of > 4. When the diagnostic threshold was increased to a score of 6, the prevalence of hypermobility was still high, 26.1% girls and 11.5% of boys [53]. In the Milhorat study, a threshold score of > 5 was used.

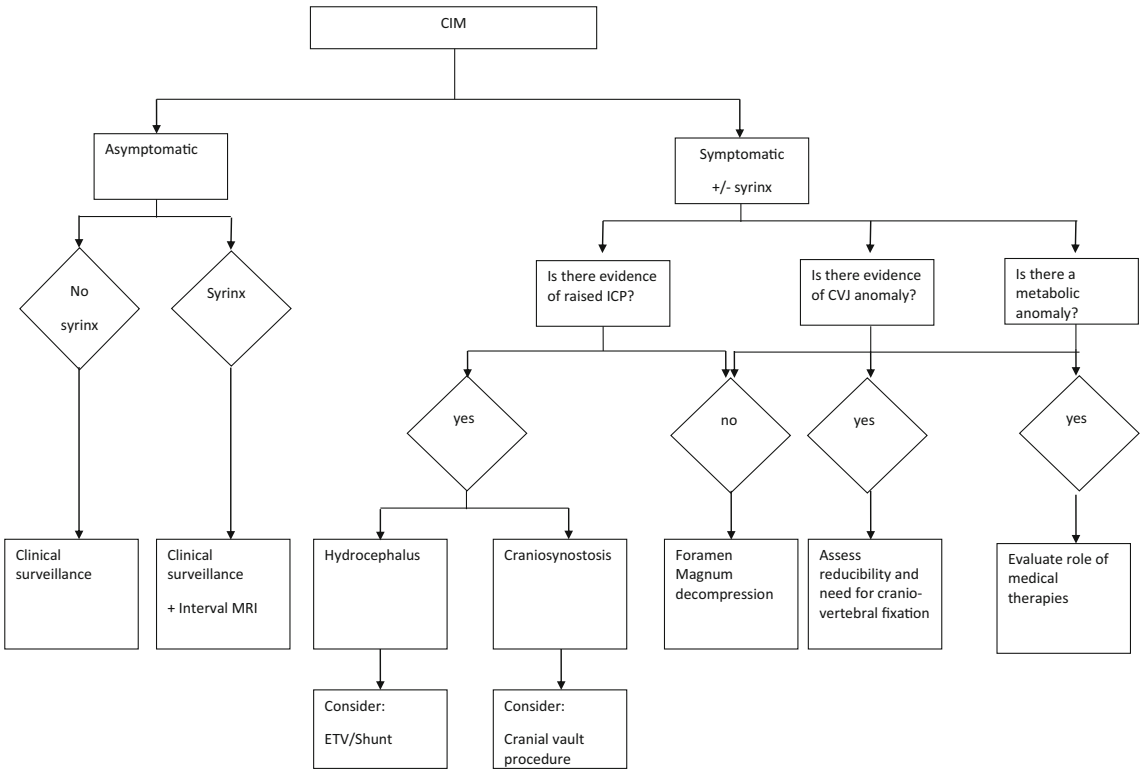


**Fig. 9** A proposed, etiology based algorithm for the evaluation and treatment of the child with CIM

As already discussed, it is well recognised that structural abnormalities of the CVJ can be an important cause of CIM, and in a proportion of patients with hypermobility disorders, with verifiable instability and CIM surgery may have a role; however, clearer diagnostic criteria and better long-term outcome studies are necessary before this can be considered a standard of care.

### Complications of Chiari I surgery—a failure to understand pathophysiology?

Favourable outcomes for headache, neurological symptoms and syringomyelia are reported in the majority of surgical series. Nonetheless, adverse outcomes are not infrequent. In an extensive literature review of Chiari treatment, encompassing 145 publications, Arnautovic et al. found that 3–5% of paediatric cases suffered a post-operative complication and 5–18% experienced a worsening of symptoms; over half of series failed to report complications [54]. Re-operation rates in the order of 16% are also not uncommon [55]. Failure to recognise underlying hydrocephalus or craniovertebral instability, along with arachnoidal scarring and dural graft related complications are among the commonest reasons for re-operation after foramen magnum decompression [22].

### Investigating Chiari I malformation

The abundance of surgical techniques and variations on the theme of foramen magnum decompression suggests that no one technique is perfect. It is perhaps our failure to acknowledge the pathophysiological diversity underlying the phenomenon of 'cerebellar tonsillar ectopia' that has led us to treat the Chiari I malformation as an isolated diagnostic entity with variable and often sub optimal results.

The evaluation of the child with CIM should begin with the premise that, far from being a congenital malformation, cerebellar tonsillar ectopia has an underlying cause and investigations should be directed to identifying the underlying disorder such that treatment can be tailored accordingly (Fig. 9).

## Conclusions

CIM remains a common reason for paediatric neurosurgical referral and its treatment has significant health economic implications. The number of publications devoted to this entity continues to rise year on year, yet numerous controversies persist. The challenge for the paediatric neurosurgeon is to try and identify those patients who may benefit from neurosurgical intervention as there are many cases of CIM that are asymptomatic, incidentally found or have symptoms unrelated to the radiological finding. Once a child is deemed to be symptomatic it is then incumbent upon the neurosurgeon to try to elucidate any underlying, predisposing pathophysiological mechanisms that have led to the CIM so that an appropriate intervention can be offered. Whilst foramen magnum decompression has been and remains the mainstay of surgical treatment for CIM, this might not be appropriate for all patients and indeed can be deleterious in some.

It is hoped that this special edition of Childs Nervous System provides a platform to acknowledge our current understanding of this condition and, perhaps more importantly, serves as a reminder of the diversity of opinion that continues to exist and the limitations of that understanding.

**Acknowledgements** This article is an abridged version dealing with aetiology of Chiari I malformation. For a more extensive review of current Chiari I management including a review of investigation and treatment, see Thompson D Chiari I Malformation and Associated syringomyelia In Textbook of Paediatric Neurosurgery Eds. DiRocco C, Pang D, Rutka J Springer 2019 ISBN 978-3-319-72167-5.

## Compliance with ethical standards

**Conflict of interest** The author declares that he has no conflict of interest.

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