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Diagnosis of Chiari I malformation and related syringomyelia: radiological and neurophysiological studies

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Abstract *Introduction:* The diagnosis of Chiari I malformation relies mainly on the anatomical demonstration of the abnormal position of the cerebellar tonsils below the foramen magnum, and of the associated bony anomalies of the posterior cranial fossa and craniocervical junction, as well as of the eventually-associated spinal cord cavitations. Consequently, the neuroradiological work-up plays a fundamental role in the definition of the malformation and in the follow-up of operated patients. *Review:* The authors review the pertinent literature on the neuroradiology of the Chiari I malformation, with special regard to MRI, with the aim of providing the reader with an updated instrument for its diagnosis.

Keywords Chiari I malformation · Syringomyelia · Cranio-vertebral junction anomalies · MRI

Radiology

According to the definition, the diagnosis of Chiari type I malformation (CIM) is based mainly on the demonstration of an abnormal position (and shape) of the cerebellar tonsils outside the cranial cavity, associated with an obliteration of the subarachnoid spaces at the level of the foramen magnum; consequently, neuroradiology plays a fundamental role in the correct identification of the anatomical aspects that characterize this malformation. Magnetic resonance (MRI) currently represents the best imaging modality. Computed tomography (CT) (Fig. 1) and CT-assisted myelography, which were the most utilized techniques before the advent of MRI, have never been completely reliable in demonstrating CIM and its related anomalies, in particular spinal cord cavitations. Even high-resolution CT scans with metrizamide can still miss some of them. On the other hand, MRI (Figs. 2, 3) provides an excellent noninvasive means of appreciating the anatomy of this malformation and of the associated anomalies, not only as part of the preoperative work-up, but also in the follow-up of surgically treated patients and of those for whom an attendant policy has been advised. Nevertheless, CT maintains its value in demonstrating bony abnormalities at the level of the foramen magnum and the craniocervical junction (Fig. 1), which are often associated with CIM, and posterior cranial fossa abnormalities, which are observed in CIM-associated craniosynostoses.

Obviously, neuroradiological investigation should first rule out any intracranial condition responsible for increased intracranial pressure and tonsillar herniation, namely tumors [8], posterior fossa cysts, hydrocephalus, etc. It is generally accepted that cerebellar tonsils situated no more than 3 mm below the basion-opisthion line, which defines the level of foramen magnum, should still be considered normal, those below 5 mm frankly pathological, and those between 3 and 5 mm "borderline". Many reports in the literature agree on these "normality" limits [1, 2, 3, 6, 7, 15, 16] although some false positives or negatives are possible utilizing these parameters. Barkovich et al. [4], analyzing a large series of normal patients, found a 99.5% specificity (i.e., only one false positive) assuming the lowest "normal" position of the cerebellar tonsils to be 3 mm below the foramen magnum. In an attempt to improve the definition of the range of "normality," Mikulis et al. [10] evaluated the effects of age on cerebellar tonsil position, and demonstrated a physiological "ascent" of these structures with increasing age; in particular, this phenomenon presented two peaks

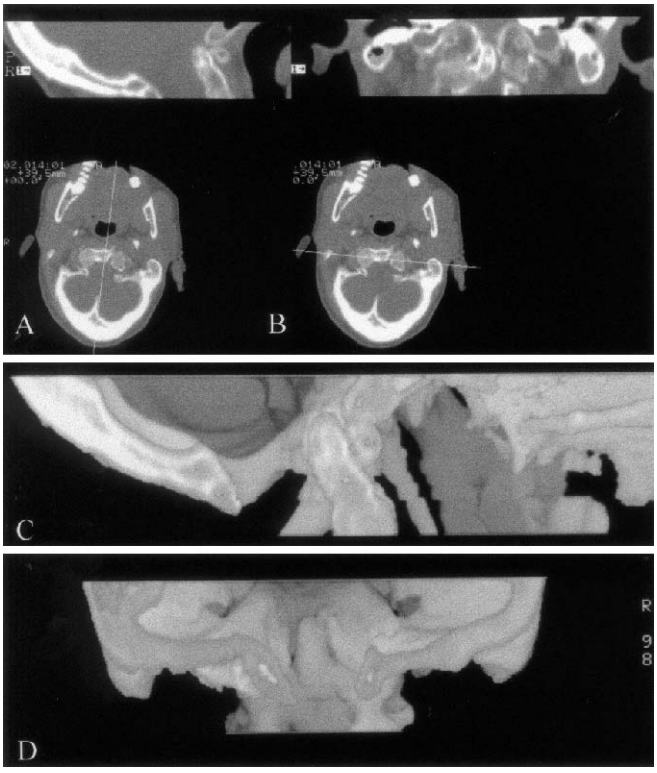

Fig. 1A–D CT scan of the craniovertebral junction of a 9-year-old boy presenting with nuchal pain and mild head tilt. Axial CT scan through the craniovertebral junction with **A** lateral and **B** frontal reformatted images. **C** Reformatted midsagittal view, and three-dimensional CT views in the **D** midsagittal plane. Note partial atlas assimilation (*right lateral mass*) and mild cerebellar tonsil displacement into the cervical canal

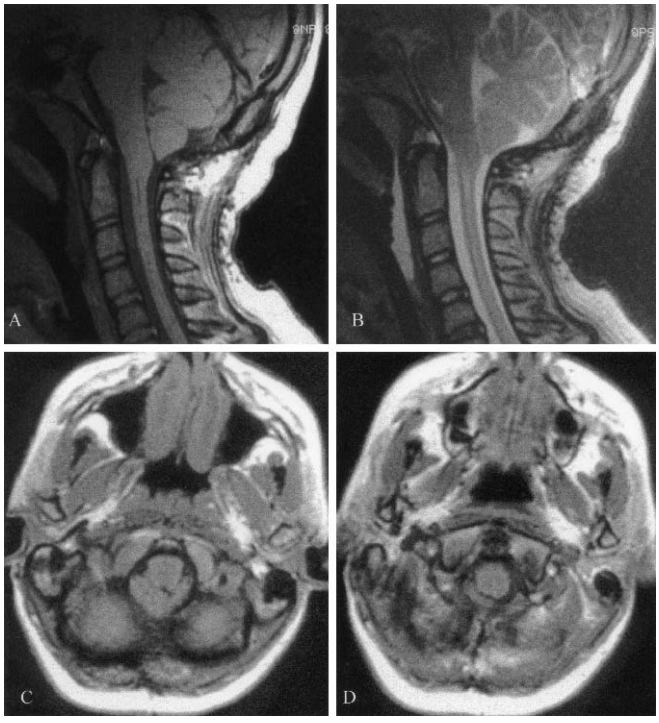

Fig. 2A–D MRI of a 10-year-old girl presenting with cervicoscapular pain. Midsagittal **A** T1-weighted and **B** T2-weighted images, and **C**, **D** axial T1-weighted images at the foramen magnum—C1 level. The cerebellar tonsils are mildly displaced into the cervical canal and their inferior aspect is pointed; the subarachnoid spaces at the foramen magnum are still present, though reduced in size. A mild increase in the size of the central canal is better appreciated on T2-weighted images (**B**)

of acceleration, one corresponding to the late childhood/adolescence period and the other to the last decades of life. A distance greater than two standard deviations beyond the normal values for the patient's age was assumed to be pathological. As an effect of this corrective factor, cerebellar tonsils lying 5 mm below the foramen magnum should be considered as pathological in late childhood and adolescence, but not in infants and toddlers for whom the pathological distance is 6 mm. In conclusion, displacement of one or both cerebellar tonsils 5 mm or more below the basion-opisthion line is the basis for the neuroradiological diagnosis of CIM; in addition, even a borderline position (3–5 mm below the foramen magnum) should be regarded as pathological when accompanied by other elements of the malformation, like syringohydromyelia and/or cervico-medullary kinking (Figs. 2, 3) [6].

In fact, as well as the position of the cerebellar tonsils, other neuroradiological aspects should be investigated to improve the definition of the severity of CIM, such as the crowding of the neural structures within the posterior cranial fossa and their impaction at the foramen magnum, the configuration of the tonsillar tips, and the coexistence of spinal cord cavitation(s). To estimate posterior cranial fossa crowding, Nishikawa et al. [13] suggested calculating the "volume ratio," defined as brain volume divided by the cranial volume of the posterior fossa (evaluated by means of MRI and CT respectively). Posterior fossa volume tends to be smaller in CIM when compared with normal individuals, even though this is not always the case. Furthermore, patients with CIM present a reduced infratentorial to supratentorial space ratio [3]. Those with a reduced posterior cranial fossa tend to present with more relevant clinical manifestations, and conversely, respond more favorably to posterior fossa decompression [7]. Since posterior fossa crowding will result in the reduction or obliteration of the subarachnoid spaces at the level of the foramen magnum, an estimation of these spaces will also be part of the preoperative work-up. Cine-MRI (see below) has been giving useful additional information on this particular aspect.

Although CIM can occur as an isolated malformation, it is frequently associated with craniovertebral junction anomalies, either congenital (like the Klippel-Feil anomaly, atlanto-occipital assimilation, platybasia, etc.) or

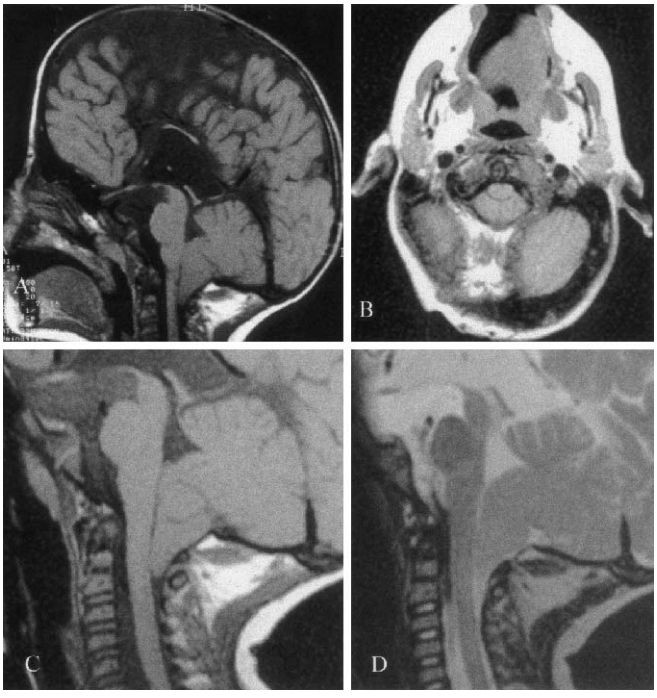

Fig. 3A–D MRI of an 18-month-old boy with syndromic craniosynostosis involving the lambdoid suture. **A** Midsagittal and **B** axial T1-weighted images, integrated by posterior fossa and cervical spine, **C** T1-weighted and **D** T2-weighted magnifications. The cerebellar tonsils are well displaced into the cervical canal and their tips present an elongated configuration; the subarachnoid spaces at the level of the foramen magnum are obliterated

acquired (like basilar invagination, craniofacial dysostosis, etc.). Foreshortening of the basiocciput results in a decreased height of the posterior fossa, with extension of the clivus into the cervical canal, crowding of neural structures, and eventual tonsillar herniation. Likewise, atlas assimilation, isolated or in association with segmentation failure of C2 and C3 (which constitutes the Klippel-Feil anomaly and occurs in almost 70% of cases), is often associated with CIM. Menezes [9] reviewing 99 patients with atlas assimilation found a CIM in 42; 32 of the latter, who also exhibited C2–C3 segmentation failure, were symptomatic. Basilar invagination, which implies prolapse of the vertebral column into the cranial cavity, is seemingly associated with CIM and syringomyelia [9, 14]. Although plain skull and spine X-rays usually allow a definite neuroradiological diagnosis, thin-section multiplanar CT with reformatted images is the best modality presently available for imaging these malformations (Fig. 1). Radiological characteristics of atlas assimilation are: (1) Partial or complete fusion of the anterior arch of the atlas with the anterior lip of the foramen magnum. (2) Lateral masses can fuse uni- or bilaterally, often asymmetrically. (3) Fusion of the posterior arch is demonstrated on lateral views as a crest overlapping on the occipital profile. The diagnosis of the Klippel-Feil anomaly relies on the radiological demonstration of the fusion of two or more cervical vertebrae, and may be difficult to image in infants and young children because the fusion site is often cartilaginous. Basilar invagination (mainly congenital) is diagnosed by demonstrating a pathological intracranial protrusion of the odontoid process at the level of the foramen magnum, whose limits are defined by means of the Chamberlain, McGregor, and McRae "lines." The exact recognition of an associated craniovertebral junction anomaly is fundamental not only for completeness of diagnosis, but also for therapeutic purposes; in fact, for instance in the case of basilar invagination, correct surgical treatment should consider ventral decompression first, before surgical decompression of the posterior fossa is undertaken. Quite recently, CIM has also been described in association with some forms of craniofacial dysostosis (Fig. 3), such as Crouzon, Carpenter, and Apert syndromes, and even in apparently nonsyndromic forms of craniosynostoses; this association has been explained to be an effect of precocious lambdoid suture fusion [5]. In particular, an elevated incidence of CIM has been reported in Crouzon and related syndromes, whereas it is much lower in the others. Rapid sequence, multiplanar, multidirectional CT fast-acquisition scans allow high quality images of cranial vault and base to be obtained in order to detect all the aspects of these malformations. Lastly, it has been reported that even an apparently "idiopathic" scoliosis can be associated with syringomyelia and CIM [12].

Concerning the appearance of the cerebellar tonsils, these may have a rounded or pointed configuration. Barkovich et al. [4] attributed these differences to the different etiology of the malformation, and suggested that tonsils tend to be large and their inferior aspect to be rounded in CIM associated with craniovertebral junction abnormalities, whereas they tend to retain a pointed aspect in cases of CIM secondary to mild intrauterine hydrocephalus. On the other hand, Iskandar and Oakes [7] interpreted a blunt and rounded aspect of the cerebellar tonsils to be an expression of a milder pathological condition, compared with a pointed and "drawn-out" configuration, which would imply a more severe condition.

Finally, neuroradiological work-up cannot be limited to the intracranial compartment but should also be extended to the spinal cord. In fact, spinal cord cavitations are demonstrated in 50–70% of patients with CIM and craniovertebral junction anomalies [9, 14, 16]; MRI is currently the best imaging modality available for diagnosing hydrosyringomyelia (Fig. 2). Both sagittal and axial T1-weighted images should be utilized, because small syringes can escape detection on sagittal cuts. They appear as low signal intensity cavitations inside the spinal cord, and isointense to CSF. On T2-weighted images a high signal intensity can be observed at the rostral and/or

caudal end of the cavity, which is attributed to microcystic or gliotic changes induced by CSF pulsation. Their more frequent location is at the cervical and dorsal levels, even though C1 is almost never affected; in about 20% of the cases the syrinx extends to the whole spinal cord ("holocord"). Multiple septations are likely to occur inside a syrinx, which however, do not usually create separate compartments.

As well as its relevance in diagnosis, neuroradiological investigation plays a fundamental role in assessing the postoperative outcome and late evolution of CIM. In fact, apart from clinical improvement, the success of posterior fossa decompression should also be integrated by neuroimaging. Unfortunately, low-lying tonsils are unlikely to ascend after surgery, and improvement in posterior fossa crowding can be difficult to assess. On the contrary, the reduction in size of a syrinx can be utilized as an objective neuroradiological sign of improvement.

Neurophysiology

As expected, brain stem auditory evoked potentials (BEAPs) should be of interest in the diagnostic work-up of patients with a Chiari malformation. Data from the literature suggest the importance of BEAPs in Chiari II malformations (an alteration of these potentials can anticipate clinical manifestations of brain stem dysfunction) and their reliability (but only when supported by a congruent clinical history and physical examination). On the contrary, their role in Chiari I is less well defined, and their contribution to surgical indication less significant. Likewise, somatosensory spinal evoked potentials (SSEPs), which are altered in the presence of even mild spinal cord dysfunction, may help with suspecting the presence of a syrinx, or may be utilized to monitor "borderline" patients for whom a "wait-and-see" policy has been advised [11]. However, according to other authors, evoked potentials should be considered unreliable in evaluating the presence and progression of hydrosyringomyelia or monitoring an apparently asymptomatic patient [6]. On the other hand, one potentially helpful neurophysiological investigation is polysomnography, which is particularly indicated in cases of children presenting with sleep apnea; in fact it can document unequivocally "central" breath alterations attributable to brain stem dysfunction, as well as their eventual improvement after posterior fossa decompression.

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