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Pathogenesis and Cerebrospinal Fluid Hydrodynamics of the Chiari I Malformation

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INTRODUCTION

The pathology that is now known as Chiari malformations of the cerebellar tonsils originated in 1891 with Hans Chiari's manuscript titled "Concerning alterations in the cerebellum resulting from cerebral hydrocephalus."[1–3] In this publication, Chiari described "alterations in the cerebellum resulting from cerebral hydrocephalus."[1–3] In 1896, Chiari described an additional mechanism for the pathogenesis of the malformation; insufficient bone growth and insufficient enlargement of portions of the skull during development cause increased intracranial pressure and subsequent tonsillar herniation.[3,4] Since Chiari's initial publications, there have been several hypotheses that attempt to elucidate the pathogenesis of the Chiari I malformation and the pathophysiology associated with it.

This article summarizes the current understanding of the pathophysiology of the Chiari malformation that is based on observations of the anatomy visualized by modern imaging with MRI and prospective studies of the physiology of patients before and after surgery. The pathogenesis of a Chiari I malformation of the cerebellar tonsils is grouped into 4 general mechanisms:

1. Overcrowding caused by underdevelopment of the posterior fossa bony structures
2. Hemodynamic disturbances that increase intracranial pressure
3. Excess tissue in the posterior fossa by a tumor
4. Downward movement of the central nervous system by events that lower intrathecal pressure, such as lumbar-to-peritoneal shunts.

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It is noteworthy that each of these mechanisms acts on normal cerebellar tonsils to deform them by impacting them in the foramen magnum, deformation that is consistently reversed by simple surgery to provide extra room at the foramen magnum.

Of note, the authors have limited their comments to Type I Chiari malformation. Even with this limitation, it becomes apparent that unanimity of thought is lacking on the pathophysiology of the type I Chiari malformation. In fact, some hypotheses even contradict each other. For example, hydrocephalus has been proposed as both the etiologic cause and a result of the Chiari malformation.[5]

PATHOGENESIS OF THE CHIARI I MALFORMATION

Limited Embryologic Development of the Skull Base

Several studies have demonstrated that many, but not all, patients with a Chiari I have a small posterior fossa. In 1 study, to investigate overcrowding in the posterior cranial fossa as the pathogenesis of Chiari malformation, Nishikawa and colleagues[6] correlated the morphology of the brainstem and cerebellum with the anatomy of skull base. They used X-Ray tomography to measure 3 occipital enchondral parts: the supraocciput, exocciput, and basiocciput. They found a significant difference in the mean length of the exocciput, from the bottom of the occipital condyle to the top of the jugular tubercle, which measured 16 mm in the Chiari group compared with 20.5 mm in control patients. They further described a significant difference in the length of the supraocciput between the internal occipital protuberance and the opisthion, which measured 38.9 mm in the Chiari group and 48.1 mm in the controls. The axial length of the clivus (the basiocciput and basisphenoid) in the Chiari group was not shorter than that of the control group. On average, the Chiari group had smaller posterior fossa cranial volume (186 cc) compared with the control group (193 cc), although it was not significant, and no significant difference was found in the posterior fossa brain volume (156 cc) compared with the controls (153 cc). As in other studies, there was a significant difference in the ratio of the posterior fossa brain volume to the posterior fossa cranial volume (mean volume ratio 0.833 in the Chiari group and 0.790 in the control group).[6]

Stovner and colleagues measured skull dimensions on lateral skull radiographs in 33 adult patients with MRI-verified Chiari malformations and 40 control subjects. They found that the posterior cranial fossa was significantly smaller and shallower in Chiari patients compared with controls. For Chiari patients, there was a positive correlation between posterior cranial fossa size and cerebellar tonsillar ectopia. Because of this positive correlation, Stovner and colleagues[7] propose that an undersized posterior cranial fossa had been expanded by hindbrain herniation at an early stage in development.

Vega and colleagues[8] studied a series of 42 patients with Chiari malformation compared with 46 control subjects. Their results support the hypothesis that cerebellar tonsillar ectopia is caused by the disproportionate size between the volume of the posterior cranial fossa and the cerebellum. The authors recorded linear, angular, and posterior fossa surface area measurements on lateral skull radiographs. They used computed tomography (CT) to calculate posterior cranial fossa volume. They found that Chiari patients exhibited shorter

clival lengths, shorter Twining-opisthion distances, and shorter Chamberlain line. Also, the average size of the posterior cranial fossa was smaller in Chiari I patients compared with control.

Perhaps the most important and often ignored developmental feature that is related directly to the pathophysiology of Chiari malformation is the somatic origin of the occipital bone. Early in embryonic development, the occipital bone forms from at least 3 pairs of sclerotomes. The occipital sclerotomes, which in turn are formed from occipital somites, eventually fuse into a single structure and are incorporated into the developing cranial skeleton.[5] Marin-Padilla examined the hypothesis that primary para-axial mesodermal insufficiency (vitamin A-induced in their experimental model) can affect embryos after the closure of the neural folds and produce a Chiari I malformation. They found that mesodermal insufficiency can produce axial skeletal defects that prevent normal neural fold closure. They propose that the Chiari malformation may be secondary to primary mesodermal insufficiency during a particular embryonic stage in the closure of the neural folds. The authors tested this hypothesis with experimental models using vitamin A as a teratogen. They were able to induce the Chiari malformation in hamsters, which suggests that the condition can be caused by an error in posterior fossa development.[5]

Finally, as a result of animal breeding to produce the characteristic skull shape of King Charles Cavalier Spaniels several centuries ago, dogs were selected with a flat shape to the back of their heads. With the introduction of the use MRI in animals, this selective breeding for a small posterior fossa, was discovered to commonly be associated with Chiari I malformation and syringomyelia.[9]

The Chiari I malformation may be secondary to underdevelopment of the occipital enchondrium, possibly from underdevelopment of occipital somites. Overcrowding in the posterior cranial fossa, due to a normal-sized hindbrain in the presence of an underdeveloped occipital bone, causes cerebellar tonsillar ectopia. However, because only a portion of patients with a Chiari I malformation have a small posterior fossa, other factors can also produce a Chiari I abnormality of the cerebellar tonsils.[6]

Hemodynamic Disturbance of the Central Nervous System

At the Cleveland Clinic in the 1950s, James Gardner, in a series of reports, established that the Chiari I malformation was the most common cause of syringomyelia.[10] He proposed that the fundamental mechanism underlying the origin of the Chiari malformation is delayed opening of the membrane covering the outlet of the fourth ventricle during fetal development and transient obstructive hydrocephalus with resulting for aminal herniation of the hindbrain and cerebellar tonsils. One of the difficulties with this potential mechanism is that it would not explain the small posterior fossa that has been described by several investigators in many patients with a Chiari I malformation since Gardner’s reports.[10]

The experimental observations of Margolis and Kilham with the reovirus-induced hydrocephalus are of considerable interest for the understanding of the morphogenesis of Chiari malformation. They inoculated hamsters postnatally with type I reovirus. This virus selectively destroys the ventricular ependymal layer, causing an inflammatory response with

subsequent gliosis. The result is an obliteration of the ventricular system at strategic points, such as the aqueduct of Sylvius, producing rapid hydrocephalus followed by cerebellar herniation. The authors propose that the Chiari malformation is caused by 2 factors:

1. The rapidly growing hydrocephalic brain will occupy the entire cranial vault including the posterior cranial fossa.
2. Due to the reduced available space in the posterior cranial fossa, the cerebellum is forced to herniate into the cervical canal, because the entire cranial vault is already filled to maximal capacity.[5,11]

In the clinic, the Chiari I malformation has also been shown to be a result of hydrocephalus and with bilateral supratentorial chronic subdural hematomas with complete resolution of the Chiari I abnormality after successful treatment.[12–14]

Herniation of the Cerebellar Tonsils Associated with Posterior Fossa Tumors

It has long been known that the mass effect of a posterior fossa tumor can produce herniation of the cerebellar tonsils with a shape that is identical to the shape of the tonsils associated with idiopathic Chiari I malformation. In 1 study, tonsil herniation identical to Chiari I malformation was evident on MRI in 24 of 164 patients with posterior fossa tumors, at least five of whom also had syringomyelia.[15]

Cranio-Spinal Pressure Imbalance Associated with Low Spinal Intrathecal Pressure

Reduced spinal intrathecal pressure with downward herniation of the cerebellar tonsils as a result of a cranio-spinal pressure imbalance caused by low spinal intrathecal pressure from a lumbar-to-peritoneal shunt or a spinal CSF leak has been known since shortly after the introduction of lumbar-to-peritoneal shunting procedures, and its presence is now considered the rule, rather than the exception, with lumbar-to-peritoneal shunting.[16,17]

Complete Reversal of the Abnormal Shape and Position of the Chiari I Malformation with Surgical Treatment of Syringomyelia

The conclusion that the typical abnormal shape and position of the cerebellar tonsils is acquired, rather than congenital, is clearly shown by complete reversion to a normal position and shape of the cerebellar tonsils in the months after successful surgery.[18]

PATHOPHYSIOLOGY ASSOCIATED WITH CHIARI I MALFORMATION

Prospective studies to examine the pathophysiology of Chiari I malformation were performed at the National Institute of Health (NIH). The NIH studies used techniques to assess anatomy, physiology with direct pressure measurements, and dynamic imaging of the tonsils with MRI and intraoperative ultrasound. This permitted simultaneous assessment and correlation of anatomic changes during the cardiac cycle with abnormal pressures. The findings of those studies have been detailed in a series of publications.[19–21] The findings of the abnormal pathophysiology have been consistent over the course of several studies, including patients with syringomyelia caused by a Chiari I malformation, patients with persistent or recurrent syringomyelia after unsuccessful surgery, and patients with primary spinal syringomyelia.[22]

Those studies indicate that a Chiari I malformation partially obstructs the free movement of CSF across the foramen magnum. In normal subjects, it is known that as cardiac systole rapidly delivers blood to the brain, the capacitance of the venous system of the brain absorbs much of the new volume of blood arriving at the intracranial space. However, during systole, about 0.75 to 1.0 mL of CSF are also rapidly moved from the cisterna magna across the subarachnoid space and into the spinal subarachnoid space at the level of the foramen magnum. That extra amount of CSF that is now in the spinal intrathecal system then moves back to the cranial space during diastole.

With a Chiari I malformation, the tonsils are impacted into the foramen magnum, blocking the rapid movement of CSF that normally occurs across that space during systole. That occlusion partially entraps the spinal intrathecal space, resulting in reduced compliance in the spinal CSF space. Now, rather than the CSF moving in and out of the head during the cardiac cycle, the tonsils rapidly move downward to provide accommodation that would normally be provided by the CSF, leaving the head and moving into the spine. That tonsil movement is against a partially entrapped CSF space with reduced compliance. In this abnormal circumstance, the tonsils act as a piston on the partially entrapped spinal CSF space to produce increased intrathecal pressure and increased pulse pressure. All these features of pathophysiology were established in the series of prospective clinical studies of Chiari I malformation at the NIH in patients with Chiari I malformation, with or without syringomyelia.[19–21]

SUMMARY

It is clear from many lines of evidence and from a broad spectrum of different circumstances that the abnormal shape of the cerebellar tonsils with a Chiari I malformation is simply a shape that is imposed on the tonsils by their impaction in the foramen magnum. This impaction occurs during the systolic delivery of blood to the brain, which occurs 100,000 to 120,000 times each day. Thus, any process that produces tonsil herniation through the foramen magnum, whether it is a small posterior fossa, herniation produced by hydrocephalous or a posterior fossa tumor, or low intrathecal CSF pressure, has the potential of producing the anatomic abnormality that is commonly called a Chiari I abnormality. The pathophysiology resulting from that impaction is simply obstruction of the free pulsatile movement of CSF across the foramen magnum, which produces the suboccipital headaches that patients experience, as well as the cough headaches and the pathophysiology underlying syringomyelia affecting some patients.

References

1. Chiari H. Über Veranderungen des Kleinhirns in Folge von Hydrocephalie des Grosshirns. Dtsch Med Wschr. 1891; 17:1171–5.
2. John A, Anson EC, Issam AA. Syringomyelia and the chiari malformations. Neurosurgical topics. AANS Publications Committee. J Bone Joint Surg Am. 1997; 80(2):308.
3. Bejjani GK. Definition of the adult Chiari malformation: a brief historical overview. Neurosurg Focus. 2001; 11(1):E1.
4. Chiari H. Uber die Veranderungen des Kleinhirns, der Pons under Medulla oblongata in Folge von congenitaler Hydrocephalie des Grosshirns. Denkschr Akad Wiss Wien. 1896; 63:71–116.

5. Marin-Padilla M, Marin-Padilla TM. Morphogenesis of experimentally induced Arnold–Chiari malformation. J Neurol Sci. 1981; 50(1):29–55. [PubMed: 7229658]
6. Nishikawa M, Sakamoto H, Hakuba A, et al. Pathogenesis of Chiari malformation: a morphometric study of the posterior cranial fossa. J Neurosurg. 1997; 86(1):40–7. [PubMed: 8988080]
7. Stovner LJ, Bergan U, Nilsen G, et al. Posterior cranial fossa dimensions in the Chiari I malformation: relation to pathogenesis and clinical presentation. Neuroradiology. 1993; 35(2):113–8. [PubMed: 8433785]
8. Vega A, Quintana F, Berciano J. Basichondrocranium anomalies in adult Chiari type I malformation: a morphometric study. J Neurol Sci. 1990; 99(2–3):137–45. [PubMed: 2086722]
9. Rusbridge C, Knowler SP. Inheritance of occipital bone hypoplasia (Chiari Type I Malformation) in Cavalier King Charles Spaniels. J Vet Intern Med. 2004; 18:673–8. [PubMed: 15515584]
10. Gardner WJ, Goodall RJ. The surgical treatment of Arnold-Chiari malformation in adults; an explanation of its mechanism and importance of encephalography in diagnosis. J Neurosurg. 1950; 7(3):199–206. [PubMed: 15415776]
11. Margolis G, Kilham L. Experimental virus-induced hydrocephalus. Relation to pathogenesis of the Arnold-Chiari malformation. J Neurosurg. 1969; 31(1):1–9. [PubMed: 4307542]
12. Di Rocco C, Frassanito P, Massimi L, et al. Hydrocephalus and Chiari type I malformation. Childs Nerv Syst. 2011; 27(10):1653–64. [PubMed: 21928030]
13. Morioka T, Shono T, Nishio S, et al. Acquired Chiari I malformation and syringomyelia associated with bilateral chronic subdural hematoma. Case report. J Neurosurg. 1995; 83(3):556–8. [PubMed: 7666236]
14. Wen L, Ma C, Wang H, et al. The role of hydrocephalus in the development of Chiari I malformation and syringomyelia. J Neurol Sci. 2014; 344(1–2):240–2. [PubMed: 25015845]
15. Tachibana S, Harada K, Abe T, et al. Syringomyelia secondary to tonsillar herniation caused by posterior fossa tumors. Surg Neurol. 1995; 43(5):470–5. discussion: 475–7. [PubMed: 7660286]
16. Welch K, Shillito J, Strand R, et al. Chiari I “malformations”—an acquired disorder? J Neurosurg. 1981; 55(4):604–9. [PubMed: 7277007]
17. Chumas PD, Armstrong DC, Drake JM, et al. Tonsillar herniation: the rule rather than the exception after lumboperitoneal shunting in the pediatric population. J Neurosurg. 1993; 78(4): 568–73. [PubMed: 8450330]
18. Heiss JD, Suffredini G, Bakhtian KD, et al. Normalization of hindbrain morphology after decompression of Chiari malformation Type I. J Neurosurg. 2012; 117(5):942–6. [PubMed: 22978540]
19. Heiss J, Suffredini G, Smith R. Comment: pathophysiology of persistent syringomyelia after decompressive craniocervical surgery. Clinical article. J Neurosurg Spine. 2010; 13(6):729–42. [PubMed: 21121751]
20. Heiss JD, Patronas N, DeVroom HL, et al. Elucidating the pathophysiology of syringomyelia. J Neurosurg. 1999; 91(4):553–62. [PubMed: 10507374]
21. Oldfield EH, Muraszko K, Shawker TH, et al. Pathophysiology of syringomyelia associated with Chiari I malformation of the cerebellar tonsils. Implications for diagnosis and treatment. J Neurosurg. 1994; 80(1):3–15. [PubMed: 8271018]
22. Heiss JD, Snyder K, Peterson MM, et al. Pathophysiology of primary spinal syringomyelia. J Neurosurg Spine. 2012; 17(5):367–80. [PubMed: 22958075]

KEY POINTS

- The pathogenesis of an anatomic Chiari I malformation can occur with several different mechanisms, including overcrowding from underdevelopment of the posterior fossa bony structures, hemodynamic disturbances of the central nervous system, such as hydrocephalus or bilateral chronic subdural hematomas producing tonsillar herniation, a mass in the posterior fossa causing tonsillar herniation, or, rarely, reduced spinal intrathecal pressure with downward herniation of the cerebellar tonsils associated with low spinal intrathecal pressure from a lumbar-to-peritoneal shunt or a spinal cerebrospinal fluid (CSF) leak.
- Because the abnormal shape and position of the cerebellar tonsils is reversed by surgery that provides unobstructed pulsatile movement of CSF across the foramen magnum, the pathogenesis of a Chiari I malformation is impaction of the tonsils in the foramen magnum, not a result of a congenital brain malformation.
- Evidence provided by anatomic imaging, dynamic imaging with MRI and intraoperative ultrasound, and physiologic studies during, before, and after surgery for Chiari I malformation is consistent with an extramedullary hydrodynamic mechanism in which the cerebellar tonsils are impacted in the foramen magnum and act on a partially entrapped spinal CSF space to increase intrathecal pressure and pulse pressure and produce suboccipital headache, and, in some patients, syringomyelia.
- Thus, the pathophysiology of the Chiari I malformation is simply the obstruction of the normal pulsatile movement of CSF across the foramen magnum.