

Factsheets for Docs-on-the-go: Chiari-1 Malformation

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You know how to recognize multiple sclerosis (MS) – you probably have several cases in your busy practice. You also expect to add one or two new cases every year, because you know that 400,000 Americans have MS, and that another 200 are diagnosed every week.¹ MS is rare, but not uncommon, so it doesn't surprise you.

Did you know that similar numbers apply to Chiari-1 Malformation? ^{2 3}

How many Chiarians have you identified in your practice?

Most cases of Chiari are left undiagnosed, or worse, misdiagnosed as multiple sclerosis, migraine, fibromyalgia, or psychiatric disturbances, a common error that prevents physically ill Chiari patients from getting the kind of help they need and deserve.⁴

Let's take a closer look.

Chiari-1 Malformation is properly defined as a mesodermal anomaly, a deformity of the posterior cranial fossa boundaries, most notably the occipital bone, but also the tentorium, clivus, basion and opisthion. The deformity subtly compresses the cerebellum and often the occipital lobe, brainstem, and/or spinal cord, too. The cerebellar tonsils herniate into the foramen magnum and may occupy a small portion of the upper cervical spinal canal. They are visible on mid-sagittal and slightly parasagittal MRIs. However, in some cases, the tonsils wrap around the brainstem or curl up like tiny snails; then their appearance is more subtle, but their effect is just as debilitating. The reason for symptom and sign manifestation – and an important key to the severity of a case – is the deformity's effect on cerebrospinal fluid (CSF) dynamics.^{5 6}

When CSF flow is turbulent, reduced, or absent in one or more locations, adjacent brain tissue receives little nutritional support, cellular byproduct removal, or cushioning from the effects of mechanical or Valsalva maneuvers. Brain function in the affected areas is disturbed. Naturally, patients report debilitating effects that correspond to the malfunction of the affected brain parts. Symptoms and signs can number into the dozens,^{7 8} can appear to exist in unrelated systems, and can wax and wane as influential factors change.

How can you identify your Chiarians?

(Yes, you do have some.) The easiest way to identify Chiarians is by cataloguing their complaints in categories corresponding to brain anatomy⁹: listen for details indicating cerebellar, occipital lobe and brainstem malfunction, then add upper spinal cord symptoms, and the malfunction of midline structures such as the hypothalamus, amygdala, and pituitary gland.¹⁰ You'll see a pattern that is unique to the patient's anatomy; it matches the territory of the malformation and reflects defects in the patterns of cerebrospinal fluid flow. Order MRI scans to verify your clinical observations.

An MRI of the brain will show subtle structural defects in addition to any obvious tonsillar herniation. An MRI of the entire spine will rule out or confirm a syrinx; this fluid-filled cavity often accompanies Chiari and can be found anywhere in the spinal cord. A Ciné MRI of the cranio-cervical junction will outline CSF flow dynamics. Watch for diminished or absent flow, especially in the retrocerebellar spaces, and from the fourth ventricle between the brainstem and cerebellum. Hydrocephalus is not necessary for the diagnosis of Chiari.

Keep in mind that subtle deformities within the posterior fossa, with or without frank herniation, can be more problematic in their effect on brain function than a large but simple ectopia that happens to leave room for normal fluid flow. This explains why short, thick cerebellar tonsils that barely plug the foramen magnum can cause a serious impediment, while a longer herniation that is thin or peg-shaped sometimes can cause few problems.¹⁰

Ok, so you realize you have a few Chiarians. Now what?

Take good care of them. Typically they have been suffering for a long time, with no name for their assailant. Explain Chiari to your patients, and reassure them that they are not psychologically ill, that their compressed brain tissue and altered fluid dynamics do have measurable effects on their physical function. Encourage them to learn more. Then work as a team to decide which of several courses you'll follow:

- 1) In cases with mild symptoms, encourage your patients to keep you informed of any changes, follow good health habits, and avoid physical activities that promote whiplash, such as trampolines, roller coasters, and contact sports;
- 2) In cases with moderate symptoms, also consider trying a biochemical treatment such as Diamox to reduce fluid production if indicated, or suggest a mechanical treatment such as cervical traction to increase space in the cranio-cervical junction;
- 3) In cases with moderate to severe symptoms, do all of the above and encourage your patients to consult with a neurosurgeon who is very experienced with Chiari cases. Surgery for this debilitating malformation is a delicate art form; it requires a resculpting of the retrocerebellar spaces to create the missing cisterna magna and to redirect cerebrospinal fluid and establish proper flow dynamics in multiple planes.

Two more points to consider:

1: Instruct your patients to report immediately any of the red-flag warning signs of severe brainstem compression: breathing difficulties, swallowing/choking problems, wild heart rate fluctuations, and/or drop attacks. When they call you, take action. Brainstem compression from Chiari can develop at a variable pace, and it has been known to kill.

2: Familiarize yourself with the subtleties of this rare-but-common disease. Check out the following internet resources for accurate, up-to-date information:

www.thechiariinstitute.com
www.conquerchiari.org
www.asap.org
www.wacma.com

1. <http://www.nationalmssociety.org/Who%20gets%20MS.asp> "Approximately 400,000 Americans acknowledge having MS, and every week about 200 people are diagnosed. Worldwide, MS may affect 2.5 million individuals."
2. <http://www.thechiariinstitute.com> "Until recent years, CM1 was regarded as a rare condition. With the increased availability of magnetic resonance imaging, the number of reported cases has risen sharply. Current estimates range from 200,000 to 2 million Americans with the condition. Genetic studies spearheaded by Dr. Milhorat support a hereditary tendency with a transmissibility rate of 12 percent. Women are affected three times more often than men. Approximately 3,500 Chiari operations are performed each year in the United States."
3. <http://www.conquerchiari.org/FAQ.htm> "Estimates for the number of people with true Chiari range as high as 500,000 in the United States. A more conservative estimate of 300,000 would mean that 1 in 1,000 people have Chiari, or 0.1% of the population."
4. Milhorat TH, Chou MW, Trinidad EM, Kula RW, Mandell M, Wolpert C, Speer, MC. Neurosurgery. 1999 May;44(5):1005-17. Chiari I malformation redefined: clinical and radiographic findings for 364 symptomatic patients.
5. McGirt ML, Nimjee SM, Floyd J, Bulsara KR, George TM Neurosurgery. 2005 Apr; 56(4):716-21 Correlation of cerebrospinal fluid flow dynamics and headache in Chiari I malformation. "Regardless of the degree of tonsillar ectopia, occipital headaches were strongly associated with hindbrain CSF flow abnormalities."
6. Quigley MF, Iskandar B, Quigley ME, Nicosia M, Haughton V. Radiology. 2004 Jul;232(1):229-36. Epub 2004 May 20. Cerebrospinal fluid flow in foramen magnum: temporal and spatial patterns at MR imaging in volunteers and in patients with Chiari I malformation. "CSF flow in symptomatic patients with Chiari I malformation, unlike that in volunteer subjects, is characterized by flow jets, regions with a preponderance of flow in one direction, and synchronous bidirectional flow."
7. http://www.thechiariinstitute.com/chiari_about.html "The majority of patients complain of severe headache and neck pain. Other common symptoms are dizziness, vertigo, disequilibrium, visual disturbances, ringing in the ears, difficulty swallowing, palpitations, sleep apnea, muscle weakness, impaired fine motor skills, chronic fatigue and painful tingling of the hands and feet. Because of this complex symptomatology, patients with CM1 are frequently misdiagnosed."
8. <http://www.asap.org/chiari-malformation.html> "Headaches are often accentuated by coughing, sneezing or straining. Patients may complain of dizziness, vertigo, disequilibrium, muscle weakness or balance problems. Often fine motor skills and hand coordination will be affected. Vision problems can also occur. Some patients experience blurred or double vision, difficulty in tracking objects or a hypersensitivity to bright lights. Physical examination may reveal nystagmus (involuntary eye movements). Other symptoms include tinnitus (buzzing or ringing in the ear), hearing loss or vocal cord paralysis. Patients may have difficulty swallowing, frequent gagging and choking and, in some cases, sleep apnea may be present."
9. <http://www.asap.org/chiari-malformation.html> "The most frequent symptom groupings include: headache, pain at base of skull/upper neck, progressive scoliosis (curvature of the spine); cerebellar dysfunction (difficulty with balance, coordination, dysequilibrium, low muscle tone); compression of the lower brainstem to cause alteration of voice, frequent respiratory tract infections, coughing when swallowing foods and fluids, compression of the spinal cord or distention due to accumulating fluid (hydromyelia); suspended alteration of sensation (e.g., arms are effected but legs not); central cord disturbance (injury to central part of spinal cord with resultant weakness greater in arms than legs); spasticity (abnormally high muscle tone or tightness, especially with movement of the muscle). Combinations of these syndromes occur commonly."
10. Milhorat TH, Chou MW, Trinidad EM, Kula RW, Mandell M, Wolpert C, Speer, MC. Neurosurgery. 1999 May;44(5):1005-17. Chiari I malformation redefined: clinical and radiographic findings for 364 symptomatic patients.

Notes

Possible Chiarians	Reasons Suspected	To Do

Additional Notes:

Thank you for making your Chiarians safer. Good luck with your busy practice. ~ SR & DW