CASE REPORT

Catching the diagnosis: A peculiar presentation of Chiari malformation type I

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ABSTRACT

Chiari I malformation is a congenital anatomic anomaly of the cerebellar tonsils resulting in their downward displacement through the foramen magnum. While often incidentally discovered on imaging with no attributable symptoms, the most common symptomatic presentation is non-specific headache. Herein, we describe a case of symptomatic Chiari I malformation in a woman with psychiatric comorbidities manifesting as a sensation of brain catching. While a peculiar description easily misconstrued by her mental health history, clinicians should consider this diagnosis in those describing symptoms compatible with headaches or occiput pain related to meningeal irritation.

KEYWORDS: Chiari I malformation; Headache; Mental health; Adult; Diagnosis; MRI

INTRODUCTION

Chiari I malformation is a congenital anatomic anomaly of the cerebellar tonsils resulting in their downward displacement through the foramen magnum. While initially considered a disease of adolescents and adults, with the advancement of magnetic resonance imaging (MRI) technology, it is being recognized at earlier stages in childhood. As such, the most widely reported prevalence of 1% is likely an underestimate [1].

Chiari I is the most common of the four Chiari malformation groups and can be associated with certain genetic syndromes, including Noonan syndrome and neurofibromatosis type 1 [2]. While most patients are asymptomatic, when symptoms are present, headaches and posterior neck pain predominate. The character of the pain and headaches are variable and can easily be mistaken for chronic tension or migraine headaches. However, if present, exacerbation with Valsalva maneuvers (ex. coughing, laughing, sneezing; all transiently increase intracranial pressure) can help clue into the diagnosis. Less common symptoms at presentation include manifestations related to compression of the herniated structures (ex. ataxia with cerebellar herniation, dysphagia with brainstem compression) [3].

Diagnosis requires imaging, with the finding of $\geqslant$5 mm of displacement of the cerebellar tonsil(s) below the foramen magnum being definitive, and 3-5 mm with syringomyelia or craniocervical junction abnormalities being highly suggestive [4]. MRI is preferred but high-resolution computed tomography (CT) can be used instead if there are system or patient factors that affect acquiring the former.

In this report, we share the case of a 20-year-old woman with psychiatric comorbidities presenting with "popping" head pain, ultimately diagnosed with Chiari I malformation. Unique to this case was the description of her presenting symptomatology and its effect on her ability to receive an accurate diagnosis when interpreted in conjunction with her mental health history. While achieving diagnostic excellence is challenging, many groups suffer from additional bias that affects their diagnostic journey and interactions with the healthcare system, including women and those with mental illness [5]. Intriguingly, there have been some reports of psychiatric comorbidities resolving with treatment of Chiari malformation. However, currently there is no definitive evidence of causality [6].

Herein, we will reinforce the epidemiology and clinical presentation of Chiari I malformation via the case as well as share the basic tenets of management. We will end by again reflecting on how mental illness can bias the diagnostic process.

CASE DESCRIPTION

A 20-year-old woman with chronic headaches, bipolar 2 disorder, and generalized anxiety disorder presented to her primary care physician with concern regarding episodic head pain. She reported experiencing sharp, shooting pain in the right posterior occiput that was accompanied by a popping sensation as if her brain was getting caught on a ledge. The symptoms had begun nine months prior, were becoming more frequent, and were associated with a near constant feeling of tension in the back of her neck. There was

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no preceding head trauma. There was no clear exacerbation of symptoms with body position or head movement though she did notice more intense symptoms on days in which she was struggling more with her mood. She had no weakness, numbness, or discoordination of her upper extremities. She did not have any involuntary movements, gait imbalance, dysphagia, or change in bowel or bladder habits.

While feeling more comfortable in her current care setting, she reported delaying presentation for these symptoms due to prior poor experiences when engaging with primary care. She was followed by a neurologist for her chronic headaches – migraine and tension type - and her symptoms had been stable for some time. Her current symptoms were inconsistent with the semiology of her headaches, present or past. She had begun treatment for her mental health eighteen months prior and was following closely with her psychiatrist. While her symptoms had improved, she reported continued struggles with self-harm, including cutting, as well as occasional use of oral psychedelics. There was no intravenous substance use. Her medications included bupropion, cyclobenzaprine, fluoxetine, sumatriptan, naproxen, and quetiapine. Her family history was significant for Sjogren syndrome and depression in her mother. She was a full-time student and was sexually active with one male partner.

Physical exam revealed normal vital signs and BMI. The right posterior scalp was mildly tender to palpation at the occipital knob without mass or deformity. Neck range of motion was normal. Neurological exam showed no focal deficits. Affect, behavior, speech, thought content, insight, and judgment were normal. Recent routine labs, including a complete blood count and complete metabolic panel, were normal. Magnetic resonance imaging (MRI) of the brain and cervical spine revealed 1.5cm of inferior tonsillar ectopia as well as diffuse T2 hyperintense fluid signal within the central spinal cord through the upper thoracic spine (Figure 1). These findings were compatible with Chiari type I malformation complicated by syrinx formation. She was referred to neurosurgery and seen a few months later. At that time, she reported new, intermittent extremity paresthesias as well as exacerbation of her symptoms with coughing and sneezing. As such, she was felt to have symptomatic disease and surgical intervention was discussed. Additional details on her outcome are unavailable due to a change in her care team.

DISCUSSION

This case highlights the importance of recognizing fewer common causes of headache given the physical and mental burden to patients that can come with a delay in diagnosis. Additionally, it highlights the power that clinicians have in framing and subsequently working through a case in an objective manner, even when the initial details and background can be a minefield for cognitive biases.

The pathogenesis of Chiari malformations remains controversial. Multiple theories exist, though none completely explain all features of the disease. The molecular genetic theory postulates the malformation results from primary defects of hindbrain segmentation and growth of associated bone and cranial structures. Another theory proposes that collision between caudally directed cranial growth and rostrally directed cervical growth is the driver. Most Chiari malformations are sporadic and not inherited, thus spontaneous mutations or deletions, or a mutation induced by an exogenous teratogen, may be implicated. The crowding theory suggests that restricted growth of the posterior fossa causes compression of neural tissue, which is then squeezed through the foramen magnum. The hydrodynamic pulsion theory suggests that early progressive fetal hydrocephalus pushes down on the brainstem and cerebellum. And finally, the oligo-cerebrospinal fluid theory proposes that defective closure of the neural tube in early fetal development results in leakage of cerebrospinal fluid (CSF) and thus insufficient cerebrospinal volume to fully distend the embryonic ventricular system, which leads to a small posterior fossa and cerebral disorganization [1,3].

The diagnosis of Chiari I malformation in adolescents and adults is made by MRI when one or both cerebellar tonsils are displaced by $\geqslant 5$ mm below the foramen magnum. Borderline displacement of the cerebellar tonsils ($\geqslant 3$ to <5 mm below the foramen magnum) is considered pathologic

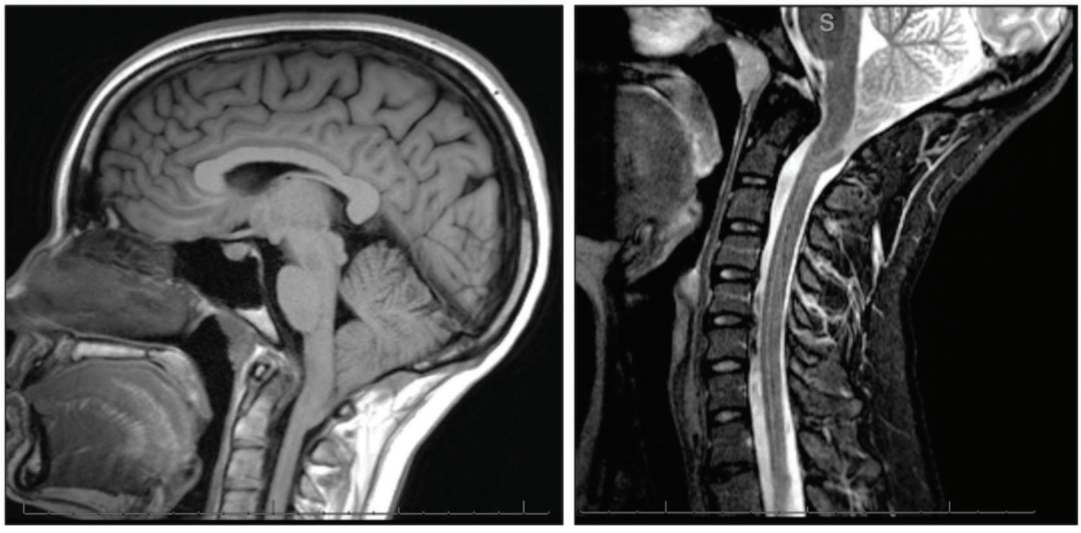

Fig. 1. Magnetic resonance imaging of brain (*left*) and cervical spine (*right*) demonstrating Chiari type I malformation.

if it is associated with additional features of Chiari I malformation, such as other craniocervical junction anomalies or syringomyelia. MRI of the brain and the whole spinal cord is the best imaging modality for evaluation of Chiari malformations. For patients who cannot have the gold-standard MRI, high-resolution CT scan with sagittal reconstructions can be used to make the diagnosis [4].

Despite a prevalence of at least 1% in the general population, the frequency of asymptomatic disease and non-specific symptoms can make Chiari type I malformations difficult to diagnose. Though worsening symptoms with Valsalva can provide a clue, its absence has inadequate sensitivity to exclude the diagnosis. Headaches are a common presenting complaint, but require comprehensive history taking and physical examination to exclude a secondary driver. Associated symptoms and exacerbating/alleviating factors should be investigated in detail, as they may be the first clue to a secondary driver. Any signal on history, exam, and/or inadequate response to treatment for primary headache syndromes should prompt advanced imaging, preferably MRI, to rule out a secondary cause such as a Chiari malformation [1,3].

Management of Chiari type I malformations varies depending on patient symptomatology. For patients who are asymptomatic and without syringomyelia on imaging, monitoring with serial imaging and examination is recommended [3]. The management for patients with asymptomatic disease, but syringomyelia on imaging, is controversial. Cases of spontaneous resolution of tonsillar displacement or syringomyelia have been rarely reported, prompting some to argue for a period of watchful observation [7]. Evaluation for CSF flow obstruction with a phase contrast cine MRI for patients is also recommended and can provide further prognostication [7,8]. For those with partial flow obstruction and limited symptoms, serial examination and imaging is reasonable. Patients with symptomatic disease, evidence of a large syrinx, or complete CSF flow obstruction should be evaluated for surgical decompression with neurosurgery. The goal of surgery is to decompress the craniocervical junction and restore the normal flow of CSF around the foramen magnum. Posterior decompression via suboccipital craniectomy, with or without duraplasty, is most often performed. Most surgeons perform decompression with opening of the dural sac for optimal decompression. The procedure involves a limited suboccipital craniectomy, C1 laminectomy, duraplasty, and arachnoid dissection. Potential complications include pseudomeningocele formation, CSF leakage, acute postoperative hydrocephalus, and meningitis. The major potential advantage of decompression without duraplasty is avoidance of these CSF-related complications, however the available data suggests greater clinical improvement and lower rates of reoperation are seen in those that undergo decompression with duraplasty. Anterior decompression of the foramen magnum, typically via transoral odontoidectomy, is an alternative surgical approach to treating Chiari malformations. It is most often used for patients with craniovertebral junction malformations who fail posterior decompression. Anterior decompression has also been used alone or in combination with posterior decompression for patients who have pronounced ventral brain stem compression associated with a Chiari malformation. Lasty, syrinx shunt placement has been used mainly for patients with Chiari I malformations who fail posterior decompression due to progressive symptoms or syrinx enlargement [7-9].

CONCLUSION

In this case, diagnosis was likely delayed in part due to the initial absence of "classic" trigger symptoms. However, her comorbid mental illness and its role in both conscious and unconscious biases cannot be overstated. Though her description of "brain popping" was perhaps atypical, recognition and additional characterization of such descriptions should be utilized instead of outright dismissal, which frequently leads to misdiagnosis, mistrust, and a delay in care, all of which our patient encountered.

Headache, particularly in association with Valsalva-induced worsening or concurrent neurological symptoms, should prompt investigation with MRI for Chiari malformation. Introspection into cognitive biases, especially against mental illness, should be acknowledged to allow proper recognition and investigation of patients' symptoms and concerns.

Informed Consent

The patient has provided informed consent for use of the case and related images, though personal details were changed to maintain anonymity as much as possible.

Financial Interests/Conflicts

We have no conflicts of interest or financial interests to report.

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