ABSTRACT

Chiari malformations are congenital defects involving the cerebral structures in the posterior fossa. They range from asymptomatic cerebellar tonsil herniation beside the brainstem, to cerebellar aplasia. These conditions were first described in 1890 and are still considered a rare occurrence even though they are more likely under-diagnosed rather than rare. We report the case of a 44 year old woman with Chiari type I malformation and associated cervical syringomyelia.

Keywords: type I, syringomyelia, syrinx, Arnold-Chiari, headache, tonsil herniation, congenital

INTRODUCTION

Chiari malformations are congenital heterogeneous defects characterized by anomalies of the cerebellum, brainstem and occipito-cervical junction with varying degrees of downward displacement of the above-mentioned structures through the foramen magnum and into the cervical canal (1). These malformations are considered rare but advances in neuroimaging and their increasing availability has proven they are, in fact, not as rare as previously thought.

CASE PRESENTATION

We report the case of a 44-year-old woman that was admitted in our Neurology Department for suboccipital headaches. Her history is positive for smoking, duodenal ulcer and severe aortic stenosis for which she underwent surgical prosthesis placement 4 years previously. The headaches first appeared two years prior and have been present ever since with periods of improvement followed by worsening. The reason she came to the hospital was that the headaches had worsened over the previous month and had subsequently affected her capacity to carry out her day-to-day activities, including work. The pain is located mainly in the cervical area with extension over the occipital area bilaterally. She also describes slight memory impairment, fatigue and bouts of vertigo. She is the only person in her family to have this type of headache. She has tried taking medication including NSAIDs, tricyclic antidepressants and triptans without significant improvement.

During the general examination we found her to be overweight (BMI = 28) with bilateral peripheral edema. Otherwise she is in good health.

The neurological examination points to be painful upon applying pressure, but otherwise was unremarkable.

We performed a cervical spine radiograph that was normal.

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The neurological examination finds both Arnold points to be painful upon applying pressure, but otherwise was unremarkable.

We performed a cervical spine radiograph that was normal.

Taking in consideration the patient complaints and the course of the pain, we decided to perform a head MRI. Sagittal T1- and T2-weighted magnetic resonance imaging (MRI) obtained demonstrates peg-like cerebellar tonsils, which protrude through the foramen magnum over a length of 8 mm and...
associated syrinx at the C1-C3 level consistent with Chiari type 1 malformation with syringomyelia. Tonsil herniation does not appear to impede CSF flow (Fig. 1, 2, 3).

![FIGURE 1. Sagittal T1-weighted image showing peg-like cerebellar tonsils, that herniate through foramen magnum 8 mm (green arrow) and associated syrinx (black arrow).](image1)

![FIGURE 2. Coronal FLAIR image showing asymmetric cerebellar tonsil herniation through foramen magnum and associated syrinx.](image2)

After consulting with the patient, we decided to treat her conservatively with carbonic anhydrase inhibitors and to monitor the Chiari malformation at 6 months. At present, the patient’s headaches have improved significantly with the medication.

It was remarkable the fact that hyposthesia due to the cavity was not present yet.

**DISCUSSION**

Austrian pathologist Hans Chiari makes the first complete description of these malformations in 1890 after observation on several autopsy specimens. Traditionally, Chiari described four degrees of severity all involving rombencephalic derivatives. A few years later, Julius Arnold published a case report of an infant with extended herniation of the brainstem and cerebellum through the foramen magnum, suggestive for what is now known as Chiari type II. The eponym now widely used (Arnold-Chiari malformation) was established in 1907 by a couple of Dr. Arnold’s students (2).

Chiari malformations are classically classified into 4 types, from I to IV; the first three represent progressively severe displacement of the cerebellum and the brainstem through the foramen magnum and subsequent impairment of cerebrospinal fluid (CSF) flow. The most severe, type IV is used to describe cases in which cerebellar hypoplasia or aplasia is found. In time, this classification has been improved by the addition of subtypes – type 0 in which the patient has a small posterior fossa and a
small, cone-like foramen magnum but without any displacement of the cerebellum and type 1.5 which is used for patients with characteristics pertaining to both type I and type II (3).

Chiari type I is characterized by inferior cerebellar migration through the foramen magnum (basion-opisthion line) and into the cervical canal of at least 5 mm. It is frequently associated with syringomyelia. Concomitant hydrocephalus is rare.

Chiari type II, also known as Arnold-Chiari malformation, is characterized by downward migration of the cerebellum – tonsils and vermis and the forth ventricle; the brainstem appears misshapen. It is found almost always in the context of myelodysplasia and hydrocephalus. Syringomyelia is almost always found in these patients.

Chiari type III is the most rare of all Chiari malformations. It is characterized by a combination of a small posterior fossa with downward displacement of the cerebellum and the brainstem into a posterior encephalocele.

Chiari type IV is characterized by cerebellum hypoplasia or aplasia and is only historically still part of the Chiari malformations (1.3,4).

Either one of the above mentioned anomalies can be accompanied by bony defects such as atlas assimilation, atlantoaxial dislocation, Klippel-Feil anomaly, platybasia, basilar invagination or lacunar skull (1).

Chiari type I is the most frequently diagnosed abnormality of all Chiari malformations. A small hypoplastic posterior fossa, in situ crowding and compression of the neural tissue and forced transforaminal displacement of the cerebellar tonsils define it. This definition, while anatomically precise can be misleading considering the fact that tonsillar herniation can be attributed to intracranial processes that are not malformations such as hydrocephalus, intracranial space occupying lesions, prolonged lumbo-peritoneal shunting, CSF leaks, tethered cord syndrome, craniosynostosis, acromegaly, and Paget’s disease.

Milhorat et al performed posterior cranial fossa morphometric measurements on 752 patients with Chiari malformations and concluded that the size of the bony elements (posterior fossa volume, occipital bone and foramen magnum size) is diminished in Chiari malformations as opposed to hydrocephalus, intracranial mass lesions, and occipito-atlantoaxial joint instability or prolonged lumbo-peritoneal shunting (5,6).

Chiari malformations are rare diseases, but some forms are more frequently encountered, especially since access to modern imaging techniques has increased in the last decade. The latest reported prevalence rates range between 0.1-0.5% with a slight female predominance. Chiari type I malformation is the most frequent across all age groups, and Chiari type II is the most frequent in pediatric populations (4,7).

Patients with Chiari type I can be asymptomatic or minimally symptomatic. Asymptomatic patients are diagnosed after incidental discovery on imaging studies performed for matters unrelated to Chiari defects (8,9).

Symptomatic patients present symptoms that pertain to elevated intracranial pressure, cranial neuropathies, medullary compression, myelopathy, cerebellar dysfunction, pain and syringomyelia (1,4,10).

Out of the symptomatic group the most frequently reported symptom was occipital headache, with certain distinguishing features – continuous headache with suboccipital localization with episodic retro-ocular component, almost never hemianopias and accompanied by vision disturbances, acoustic or vestibular disturbances, paresthesias and signs of medullary involvement. The pain is made worse by Valsalva type maneuvers, it is sometimes worse in the premenstrual period and it rarely is sensible to antidepressants, betablockers or triptans. It can be alleviated by topiramate, which lowers CSF pressure due to its effect as a carbonic anhydrase inhibitor (5,11,12).

Neurologic examination is most often normal. Patients come seeking medical advice due to the worsening of headaches. Typical findings include vestibular-like disequilibrium and difficulty standing and walking in tandem.

Diagnosis of Chiari malformation type I is made using imaging studies, particularly MRI of the brain and cervical spine, which visualize the herniation of the cerebellar tonsils of at least 5 mm through the foramen magnum and into the central canal. Sagittal plane sequences are most useful in visualizing the posterior fossa and the occipitocervical junction. Several measurements need to be made: the diameter of the posterior fossa, the diameter of the foramen magnum (Twining line) and the size of the occipital bone. All these structures should be diminished in size in Chiari I malformations, otherwise a different cause for the herniation should be sought out. It is useful to mention that the foramen magnum is enlarged in tethered cord syndrome and Chiari type II and stenosed in Chiari type I, craniosynostosis, and achondroplasia. The sagittal and coronal planes are useful in visualizing the crowding of the posterior fossa and the foramen
Chiari malformations are not as rare as we think. They are probably more frequent but are left undiagnosed, due to lack of symptomatology. These patients have to be carefully selected before a decision for surgical treatment is made. Goals for surgery are brainstem decompression and reestablishment of normal CSF flow from the “crowded” posterior fossa and into the cervical subarachnoid space.