"Acquired" Chiari I malformation

Case report

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Tonsillar descent of the cerebellum in Chiari I malformations is often considered a congenital defect. A patient is presented in whom magnetic resonance (MR) imaging revealed normally positioned cerebellar tonsils; however, 1 year later MR imaging was repeated for evaluation of gait abnormalities and showed descent of the cerebellar tonsils. This case illustrates worsening symptoms with progressive descent of the cerebellar tonsils and suggests that Chiari I malformations can evolve postnatally.

KEY WORDS • Chiari malformation • cerebellum • tonsillar herniation

Traditionally, the Chiari I malformation is depicted as a congenital rather than an acquired defect. Chiari described Type I malformations as "elongations of the tonsils and medial part of the inferior lobes of the cerebellum which go along the medulla into the cervical canal." These malformations are associated with hydrocephaly and cavitation of the spinal cord in 50% to 75% of patients. Chiari believed that cerebral hydrocephalus caused herniation of the posterior fossa contents. Gardner restated this concept in a different form. However, most patients with Chiari I malformation do not exhibit hydrocephalus and most instances of congenital hydrocephalus are not associated with tonsillar descent. More recently, cases of "acquired" Chiari I malformation have been documented in patients with supratentorial mass lesions and after removal of cerebrospinal fluid (CSF) via a spinal shunting procedure or multiple lumbar taps.

In the following case report, seizures in a 6-month-old infant led to initial unremarkable magnetic resonance (MR) imaging. Follow-up MR imaging obtained 1 year later because of worsening gait difficulties revealed tonsillar descent. This case provides evidence that Chiari I malformations may evolve postnatally.

Case Report

This 22-month-old right-handed baby girl was the first child born as the product of a full-term, spontaneous vaginal delivery to a 28-year-old white woman. A partial placenta previa had resulted in mild spotting during the second trimester of the mother's pregnancy. A mildly elevated alpha-fetoprotein level was noted during routine perinatal testing, but results from the ensuing ultrasound study were unremarkable. The Apgar score at birth was 9/9 at 1 and 5 minutes. The patient did well until 6 months of age when she developed an upper respiratory infection. She was brought to her pediatrician and received her third diphtheria-pertussis-tetanus shot during that visit. That evening she experienced a generalized tonic-clonic seizure associated with a body temperature of 103°F. The infection and fever resolved, but the patient suffered a second generalized tonic-clonic seizure 8 days later without a fever. She continued to have generalized tonic-clonic seizures approximately once a week and to exhibit breakthrough seizures despite multiple drug regimens. Several weeks prior to admission to our hospital, the patient's medication was switched to valproic acid and nitrazepam. The generalized tonic-clonic seizures ceased, but she began to experience absence seizures while on the new regimen. At 8 months of age, an MR image was unremarkable apart from mild atrophy (Fig. 1). Multiple electroencephalographic recordings were consistent with generalized seizure activity, but did not demonstrate any focal pathology. The patient began walking at 15 months of age; however, her motor skills did not progress and she became increasingly clumsy with poor balance and lower-extremity ataxia. A developmental and metabolic workup was unremarkable, as were a spinal tap and muscle biopsy performed at 20 months of age. Magnetic resonance imaging of the spine showed the tip of the conus at the L1-2 interspace...
and revealed no tethering lesions. Eventually, MR imaging of the brain was repeated, showing a Chiari I malformation (Fig. 2) that had not been present on the previous image.

**Examination.** The patient had no history of respiratory problems, although it was reported that over the past few months she had an increasing tendency to choke during meals as well as an elevation in the pitch of her voice. On occasion, she would appear to be in some discomfort, rubbing her head and eyes as if she were having headaches. The patient verbalized sounds but did not enunciate clearly recognizable words. She interacted well with her environment, was quite attentive, and easily engaged in play.

Her physical examination was notable for an absent or atrophied pectoral muscle on the left side. She appeared to have bilateral winged scapulae, as did her father. No cutaneous markings were noted. Motor strength and sensation appeared grossly normal and symmetrical in all extremities. The patient was able to reach for objects quite smoothly with her upper extremities. Pathological reflexes could not be elicited. She could bear her own weight but would sway from side to side while walking and would fall after several steps.

**Operation.** The patient was admitted to our institution for decompressive surgery. Preoperative laboratory tests showed abnormalities in her liver and coagulation profiles, which were thought to be secondary to ingestion of valproic acid. The patient was discharged with the plan to readmit her after laboratory test values had normalized. However, several days after discharge the patient experienced progressive worsening of her symptoms, with increasing difficulty in bearing her own weight and holding her head upright. She was readmitted and underwent a C1–2 laminectomy and duroplasty. At the time of surgery, the tonsils were noted to be at the level of the C-2 lamina. Tonsillar pulsations were noted upon opening the dura, and the tonsillar tips were freely mobile. No other anatomical abnormalities were noted intraoperatively.

**Postoperative Course.** Postoperatively, the patient's symptoms stabilized. At the time of discharge, the patient was again able to bear her own weight and to hold her head upright. Her gait abnormalities, although not improved, remained stable after 4 weeks.

**Discussion**

Cerebellar ectopia is traditionally seen as a congenital finding. Chiari believed that hydrocephalus produced the tonsillar herniation seen in Type I malformations. However, hydrocephalus is not routinely associated with Type I deformities and thus cannot be the explanation in most cases of these malformations. Gardner modified this concept by suggesting that transient hydrocephalus following closure of the neural tube contributed to cerebellar herniation. There are some inconsistencies with this theory. First, the transient nature of the proposed hydrocephalus should lead to ascent of the tonsils back to their normal position. Second, fusion of the neural folds occurs on the 22nd day of gestation, with closure of the anterior neuropore at 25 days and the posterior neuropore 2 days later. Since the cerebellar hemispheres do not form until 5 months of gestation, transient downward pressure at this stage of development would not involve the still unfomed hemispheres.

Others have attributed the tonsillar herniation associated with Chiari I malformation to a disproportion between the posterior fossa and its tissue contents. In animal models, high doses of vitamin A induce a reduction in the size of the posterior fossa and caudal displacement of the cerebellum, which increases during the postnatal growth spurt. Several studies in humans suggest that a mismatch between the volume of the cerebellum and the posterior fossa may contribute to downward herniation of the cerebellar tonsils. Patients with Chiari I malformation have, on average, a smaller and shallower posterior fossa than normal individuals. However, there is a large degree of overlap between patients with Chiari I malformation and control subjects with regard to posterior fossa size. In fact, in a large proportion of Chiari I patients the posterior fossa has normal dimensions. Of interest,
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Stovner, et al., 22 showed that symptomatology was inversely related to posterior fossa size despite a positive correlation between posterior fossa size and cerebellar ectopia. This relationship was thought to be secondary to remodeling of the posterior fossa during hindbrain herniation. There is as yet no evidence that a small posterior fossa by itself is responsible for the production of neurological signs and symptoms in Chiari malformations. These data also do not explain those patients who develop symptoms in the fifth and sixth decades of life, well after growth is established.

There is much evidence that the symptomatology of Chiari I malformation is an acquired process. The Chiari I deformity rarely presents in early childhood or infancy. Despite the congenital nature of this malformation, most patients present with symptoms in the second to sixth decade of life.1,10,15 Swift and Carmel25 describe a group of patients with "transitional" Type I anomalies followed via serial MR imaging. The progressive nature of symptoms and reversal of the tonsillar herniation and medullary "kink" postoperatively in several cases again suggest that this is an acquired deformity.

Other examples of acquired Chiari I malformation have been documented.5,12,19,26 Several investigators have shown that cerebellar descent can occur during removal of CSF by lumbar shunting or by repeated lumbar taps.5,12,20,27 The tonsillar herniation resolves after removal of the shunt.25,27 Given this observation, it seems reasonable that loss of CSF via an open caudal spine and the caudally directed pressure gradient may account for some instances of tonsillar descent. Welch, et al., 27 proposed a physiological mechanism for tonsillar descent caused by differences in the CSF absorptive properties of the cranial and spinal compartments. These pressure differentials could be a potential force for the spontaneous development of a Chiari I malformation. Cerebrospinal fluid is clearly absorbed via spinal pathways.6,11,18 and recruitment of these pathways has been shown in animal models.7 One can imagine circumstances in which the normal cephalic absorptive mechanisms are impaired with maintenance of function in the spinal pathways, a condition comparable to the situation after spinal shunting.

Finally, spinal cord elongation during normal growth introduces a downward vector on the cervical spinal cord and the hindbrain, and may contribute to tonsillar descent. Such a process may even occur in the absence of pathological tethering in the lumbar sacral region.

This is the first documented case where normally positioned cerebellar tonsils descended over the span of 1 year without external intervention such as lumpectononal shunting. It has always been somewhat puzzling when a patient with a Chiari malformation presents in the fourth or fifth decade of life with new symptoms. Most authors assume that tonsillar descent is present at birth with later symptomatology secondary to either continued or increasing tonsillar impaction upon the brain stem. Although one can imagine a scenario wherein the onset of symptoms occurs only after continuous slow impaction on the brain stem over decades, our data suggest that new symptoms may develop as a result of a subacute descent of the cerebellar tonsils. Obviously, there will be a wide spectrum of examples between these two extremes. Our patient is atypical in her presentation of symptoms at a relatively young age, which may be related to the dramatic change in the position of her cerebellar tonsils over a relatively short time span.

As the use of MR imaging becomes routine, we imagine that additional examples of progressive cerebellar descent will be seen. Tonsillar descent in patients with Chiari I malformation is presumably caused by a combination of mechanical and congenital factors that together promote the downward herniation of the cerebellar tonsils. This case provides evidence that the progressive symptomatology seen in some of these patients may be the result of a dynamic rather than a static process.

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