Gait disturbances and seizure-like episodes in a patient with a Chiari malformation

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CHIARI MALFORMATIONS

Chiari malformations are a diverse group of congenital malformations that involve the cerebellum, brainstem, and cranio cervical junction in the posterior fossa.1 Chiari malformations were first described by John Cleland in 1883, but the term is used in recognition of Hans Chiari, who described a case of tonsillar ectopia in 1891.2 The term Arnold-Chiari is also used to describe such malformations with spinal dysraphism, in recognition of Julius Arnold.3 Three types of Chiari malformations have been identified to be compatible with life, each involving distinct clinical and anatomical features, but all involving various degrees of cerebellar descent through the foramen magnum.4 Chiari malformations are diagnosed on the basis of neuroanatomical findings alone, and thus magnetic resonance imaging (MRI) is the primary modality for evaluation.4 There are no tissue, blood or cerebrospinal fluid (CSF) biomarkers to confirm the diagnosis.1

The Chiari I malformation (CMI) is characterized by downward displacement of the cerebellar tonsils through the foramen magnum into the upper cervical canal.5 Tonsillar ectopia of 5 mm or greater is generally consistent with CMI. However, an association between the degree of cerebellar descent and clinical severity is unclear.6 As such, the clinical manifestations of CMI are highly variable, and may be due to the amount of nervous tissue displacement and the degree of compression.6 Primary symptoms include tensive headaches, visual changes, syncope, muscle weakness, dysarthria and ataxia.7

Chiari II (CMII) and III (CMIII) malformations are associated with neural tube closure defects, and occur much less frequently than CMI. CMII is characterized by a small posterior fossa with downward displacement of the cerebellar tonsils and inferior cerebellar vermis into the foramen magnum and upper cervical canal, in association with a myelomeningocele.1,8 These patients tend to have hydrocephalus,4 and clinical manifestations may include lower cranial nerve deficits, cerebellar dysfunction, and respiratory disturbances.9 CMIII is exceedingly rare, and is defined as cerebellar displacement into an occipital or cervical cephalocele.8 This condition has a high early mortality rate, and causes several severe neurological deficits in survivors, including delayed milestones, seizures, ataxia, and spasticity.9

CASE

LG is a 2 year old boy who presented in May 2012 with progressive gait ataxia and generalized weakness, in addition to falling down approximately six times per day. These symptoms were accompanied by slurring of speech, neck pain, and headache without nausea or vomiting.

His past medical history is significant for similar episodes, described as “grey spells,” which presented within the first few months of life. He has had multiple Emergency Department admissions for issues of falling, associated with respiratory symptoms and hypotonia. In addition, his mother would describe “shaking episodes” reminiscent of generalized tonic-clonic seizures. These symptoms were evaluated carefully: a thorough cardiac examination did not reveal any pathology, nor did an ophthalmological evaluation or a detailed genetic/metabolic screen (including blood and urine amino acids, urine organic acids, plasma ammonia, lactate, and carnitine levels, and urinary acylcarnitine profiles). Multiple EEG evaluations, including several 24-hour video EEGs revealed no evidence to support a diagnosis of clinical or electrographic seizures. A tentative diagnosis of alternating hemiplegia of childhood was made, which is a poorly understood disorder, and primarily a diagnosis of exclusion.10

MRI of the head demonstrated extension of the cerebellar tonsils 7 mm below the foramen magnum, which is consistent with CMI. At that time, it was decided to forego surgical intervention due to the moderate severity of his symptoms. However, he subsequently developed more frequent seizure-like activity without any supportive evidence of the events being epileptic in origin, and follow-up MRI demonstrated an additional extension of 4 mm. Thus, it was decided to perform a suboccipital craniecostomy for posterior fossa decompression. Unfortunately, this surgery was complicated by two subsequent CSF leaks from the initial incision, as well as bacterial meningitis secondary to the CSF leak.

Following these complications, LG experienced only a single event prior to his current admission. While eating in a restaurant, his head flopped backwards, accompanied by a transient depression of his level of consciousness and abnormal extensor posturing. A thorough assessment revealed no focal neurological deficits, unremarkable physical exam and blood work, unremarkable EEG and MRI, and negative lumbar puncture cultures.

With regards to his current admission, MRI did not demonstrate any interval change. EEG evaluation again did not reveal epileptiform activity. LG did not demonstrate any symptoms in hospital as had been reported by his family, and he was discharged home without any additional therapy or medications.

DISCUSSION

The presenting symptoms in patients with CMI are diverse and usually necessitate investigations for other pathologies before the definitive diagnosis is made. These manifestations can often be related to dysfunction of the brainstem, cranial nerves, spinal cord, or cerebellum.

The management of CMI depends on the presenting symptoms and associated neurologic impairments. It has been suggested that asymptomatic patients can be managed conservatively with serial MRI and regular clinic visits,12 although some would advocate for prophylactic surgery to prevent neurological sequela.13 The goal of decompressive surgery is to
Clinical scenarios are not mutually exclusive and can overlap with one another. At times, the presenting complaints can be vague, and the diagnosis is often delayed, which is typical in cases of Chiari I malformations. The most common presenting symptoms include cerebellar ataxia, headaches, neck pain, and sleep disorders. In some cases, patients may experience drop attacks, extensor posturing, and varying degrees of respiratory compromise and described these episodes as "cerebellar fits." This term was first used to describe tetanus-like episodes in a young boy with a midline cerebellar tumor, but has since been expanded to include patients with tonsillar herniation. However, it was also noted that decompressive surgery was sufficient to alleviate these symptoms.

Patient LG continued to be symptomatic despite surgical intervention, although the frequency of such episodes was significantly less. Data regarding surgical outcomes show variable post-operative recovery in pediatric populations. One retrospective study demonstrated symptomatic improvement in 68-72% of patients, whereas another demonstrated improvement in 83% of patients.

The continued occurrence of these grey spells at present remains unexplained as recent imaging did not show any obstructive pathology in the foramen magnum. Previous studies have suggested that such a recurrence may be due to the size of the decompression, post-operative scar-