Chiari I Malformation Causing Developmental Regression in a 4 Month Old

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Introduction

Chiari I malformation is characterized by downward displacement of cerebellar tonsils through the foramen magnum. In the era of magnetic resonance imaging (MRI), specific radiological criteria for the diagnosis of Chiari I malformation has emerged. In both children and adults, diagnosis is established by displacement of one or both tonsils 5 mm below the plane of the foramen magnum. However, Mikulis et al examined a group of 221 patients ranging from 5 months to 89 years, and found that tonsils tend to ascend with age. They recommend that criteria be modified for age, such that younger patients are permitted a higher degree of ectopia.

The most commonly reported symptom in both adults and children is headache, occurring in more than half of patients and is often precipitated by Valsalva maneuvers. Younger children tend to present more commonly with dysphagia and respiratory difficulties. Before children can communicate verbally, symptoms like headache may present with crying or irritability. Neurological signs of Chiari I malformation can be codified into 3 categories: a brainstem syndrome (eg, cranial nerve dysfunction, respiratory impairment), a spinal cord syndrome (eg, motor and sensory loss, hyperreflexia or hyporeflexia), and a cerebellar syndrome (eg, ataxia). Syringomyelia is associated with Chiari I malformation and deserves special mention as it often warrants surgical intervention.

Case Report

A term gestation twin male infant presented to the emergency department at 4 months of age with 3 weeks of decreased formula intake and decreased use of his arms. During this interval of time, he stopped rolling over, lifting his head from the prone position, or reaching for objects. Pregnancy and prior medical history were unremarkable. There was no family history of Chiari I malformation or other neurodevelopmental disorders. There is no report of trauma prior to onset of symptoms.

General physical exam was remarkable only for growth parameters at the third percentile for age, including the orbital-frontal circumference (38 cm); this was similar to his twin sibling. Neurological examination on admission showed hyperreflexia in upper and lower extremities with sustained clonus at the ankles; axial hypotonia with head lag and slip-through on vertical suspension; and normal muscle bulk but decreased apparent strength. MRI brain with and without contrast was obtained and showed descent of the cerebellar tonsils 15 mm below the plane of the foramen magnum, as well as cervicomedullary compression and edema at the C4-C5 level (Figure 1).

The patient underwent suboccipital decompression, C1 laminectomy with tonsillar reduction, and duraplasty. His immediate postoperative neurological exam showed improved spontaneous arm movements. Postoperatively the patient developed hydrocephalus at the foramen magnum level, requiring ventriculostomy, after which a brain MRI showed diffusion restriction in the bilateral cerebellum in the distribution of the posterior inferior cerebellar arteries. However, the MRI also showed decreased edema and T2 prolongation in the cervical cord.

At neurology follow-up at 7 months of age, he was only slightly behind his twin brother in terms of developmental attainment. He could transfer objects from hand-to-hand and was almost able to sit independently.
Examination revealed age-appropriate movement of his arms and legs, normal tone, and mild hyperreflexia of deep tendon reflexes throughout. At his most recent follow-up at 15 months of age, he requires physical therapy for some incoordination while walking on uneven terrain and speech therapy for mild oral aversion.

Discussion

We report an unusual cause of developmental regression related to an early presentation of Chiari I malformation. Several frameworks exist for classifying disorders causing progressive developmental regression and most focus broadly on genetic causes, including inborn errors of metabolism and neurodegenerative disorders. Inborn errors of metabolism are generally characterized by toxic accumulation of compounds or inability to synthesize certain products, whereas neurodegenerative disorders do not have a specific metabolic underpinning but are marked by progressive neuronal loss. Structural brain malformations are typically thought to be static or nonprogressive, but as this case report demonstrates, structural defects can cause progressive developmental regression.

The patient’s brain MRI showed cerebellar tonsillar descent with cervical cord edema. Although there is not a radiological definition for Chiari I in infants, the clinical presentation and imaging findings are consistent with a symptomatic Chiari I malformation.

Few studies focus exclusively on Chiari I malformations in infants, but oropharyngeal dysfunction is reported most commonly, which includes disparate symptoms such as dysphagia, reflux, respiratory difficulties, and failure to thrive. Several authors have also reported infants with motor impairments, ranging from focal weakness to quadriplegia. Motor findings are generally associated with syringomyelia, and most of these infants underwent surgical decompression with positive outcomes. This patient also presented with motor deficits, manifesting as developmental regression, and also responded well to surgical intervention.

Developmental regression is an ominous sign in an infant. We report the rare association of arm diplegia, spinal injury, and Chiari type I malformation in a 4-month-old infant and recommend that an early surgical approach should be considered in these patients. Additionally, the young age of presentation in this patient suggests an underlying genetic predisposition for development of symptomatic Chiari I malformation.

Author Contributions

Drs Doll, Morita, Bonkowsky, Brown, de Havenon, Brockmeyer, and Glasgow had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the analysis. Study concept and design: Doll, Morita, Glasgow. Analysis and interpretation of data: Doll, Morita, Bonkowsky, Brown, de Havenon, Brockmeyer, and Glasgow. Drafting of article: Doll, Morita, Bonkowsky, Brown, and de Havenon. Critical revision of the article for important intellectual content: Morita, Bonkowsky, Brockmeyer, and Glasgow.

Declaration of Conflicting Interests

The author(s) declared the following potential conflicts of interest with respect to the research, authorship, and/or publication of this article: Drs Morita and Bonkowsky receive royalties from a published book—Bale JF Jr, Bonkowsky JL, Filloux FM, Hedlund GL, Larsen PD, Nielsen DM. Pediatric Neurology. London, England: Manson; 2011.

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Ethical Approval

The study was approved by the institutional review boards of the University of Utah and Intermountain Healthcare.
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