



Progressive Flaccid Paraplegia in a Toddler due to Chiari Type I Malformation Complicated with Hydrocephalus and Syringomyelia. A Case Report

Angelina Lo, BS¹ , Megan C. LaRocca, BS¹, Danielle Whalen, MD², and Zurisadai Gonzalez-Castillo, MD² 

Abstract

Chiari malformation is a clinico-radiological entity defined by herniation of rhombencephalic structures through the foramen magnum. The most common type, Chiari I, involves herniation of the cerebellar tonsils specifically. We present the case of a 2-year-old with three weeks of progressive bilateral leg weakness, absent reflexes, and the inability to walk. The patient was found to have Chiari I with hydrocephalus and syringomyelia. This is the youngest patient reported in the literature presenting with a clinical picture of spinal shock. Early recognition of this entity allows for proper treatment and improved outcomes.

Keywords

malformation, pediatric, radiology

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Introduction

Chiari I malformation is a rare congenital malformation in which cerebellar tonsils are downwardly displaced through the foramen magnum into the cervical spinal canal. This anatomic anomaly may cause restricted growth of the cerebellum, brainstem compression, and blockage of cerebrospinal fluid (CSF) flow through the foramen magnum. Chiari I can be asymptomatic and found incidentally or symptomatic with an average presenting age of 18 years. Signs and symptoms differ in children younger than six years from those seen in older patients, and few reports exist documenting this early clinical presentation. The cerebellar obstruction may result in complications such as concurrent syringomyelia and/or hydrocephalus in up to 25% and 10% of patients, respectively.^{1,2} Other common manifestations include dysphagia, vision problems, hearing problems, difficulty sleeping, ataxia, muscle weakness, or paresthesias.

Oropharyngeal dysfunction is the most common presenting symptom in infants and toddlers under the age of two although symptomatic presentation of Chiari I malformation is rare in children under the age of six.³ To the authors' knowledge, this is the first documented case of bilateral leg weakness and

hyporeflexia secondary to syringomyelia in a toddler with Chiari I malformation.

Case

A 2-year-old boy presented to the emergency department (ED) with three weeks of bilateral leg weakness and refusal to walk which was preceded by a respiratory illness two weeks prior to initial presentation. The patient had an uncomplicated birth history, normal gross motor development, and at baseline, was able to run, walk up stairs, and kick a ball. Language development was delayed as the patient did not yet speak 50 words or in 2-word phrases. Parents initially noticed toe walking and externally rotated feet, followed by unsteady gait and multiple

¹School of Medicine, University of California, Irvine, Irvine, CA, USA

²Department of Child Neurology, Children's Hospital Orange County, Orange, CA, USA

Corresponding Author:

Angelina Lo, School of Medicine, University of California, Irvine, 1001 Health Sciences Rd, Irvine, CA, 92617, USA.

Email: aslo@hs.uci.edu



falls. Upon presentation to the ED, he was afebrile with normal vital signs. Physical exam showed shuffling gait with externally rotated feet while ambulating and no resistance to movement of lower extremities. Workup showed a normal complete blood count (CBC), comprehensive metabolic panel (CMP), negative inflammatory markers, normal bilateral lower extremity x-rays, and bilateral hip ultrasounds that showed trace right hip effusion. Given his recent history of respiratory illness, presenting symptoms, hip ultrasound findings, and improvement with NSAIDs, he was diagnosed with transient synovitis and discharged home.

He returned to the ED 2 weeks later with persistent leg weakness and refusal to stand up and walk since discharge. The patient was noted to crawl on all four extremities in the interim without any evidence of pain or arm weakness. Vitals signs were within normal limits. During his second hospitalization, neurology was consulted, and his neurological exam showed the following motor and sensory findings: the patient was lying in bed, legs in a frog-legged position, and he used his arms to move and reposition his legs. He had normal bulk and decreased appendicular tone. He was able to move both arms and resist the examiner, however lower extremity strength was two out of five bilaterally. There was normal sensation in upper extremities, but he did not withdraw to noxious stimuli applied to bilateral legs. Reflexes were 1+ in bilateral upper extremities with absent patellar and Achilles reflexes. When pulled up to stand, he was not able to bear weight on his feet without assistance.

It was recommended to obtain neuraxis imaging (Figure 1) that demonstrated crowding and inferior displacement of cerebellar tonsils 13 mm below the foramen magnum. There was dilation of the third, fourth, and lateral ventricles consistent with hydrocephalus. MRI of the C-, T-, and L-spine showed cord edema throughout the cervical spine, a syrinx from C3-4 extending inferiorly to T8-T9, and displaced peripheral parenchymal thinning extending to T8-T9. Below this level, there

was cord expansion with hyperintense T2 signals suggesting continuation of cord edema and/or syrinx to the conus.

Neurosurgery was consulted, and an urgent suboccipital decompression and C1 laminectomy was performed with no complications. The cerebellar tonsils were cauterized and shrunk to the level of the occipital bone edge with good CSF flow from the fourth ventricle down. At his follow-up visit on postoperative day 21, the patient showed improvement in strength and motor function. Though the patient was hesitant to walk on his own, he was able to stand independently. Follow-up MRIs taken 2 months after surgery (Figure 2) showed adequate decompression, resolution of hydrocephalus, and decreased size of syrinx. He is attending physical therapy and is expected to continue his recovery.

Discussion

Syringomyelia is a disorder in which a CSF-filled cyst forms within the spinal cord. It is most commonly associated with sensory loss in a cape-like distribution due to central cord compression of the cervical spine. Holocord syrinx expansion may cause compression of the thoracic, lumbar, and sacral nerve fibers, resulting in varying neurologic deficits of the trunk and lower extremities. Chiari I malformation predisposes patients to syrinx formation, as it interferes with normal CSF flow through the foramen magnum.⁴

Few reports of Chiari I presentation in children under six exist, and the signs and symptoms differ from those seen in older patients. Albert et al reported a pediatric cohort that included 31 patients and demonstrated that children aged 0–2 years were more likely to present with oropharyngeal dysfunction and those aged 3–5 years were more likely to present with scoliosis, syringomyelia, and/or headache.³

In the present case, a large holocord syrinx extended inferiorly to the thoracic and lumbar spine causing rapidly progressive leg weakness and hyporeflexia. To the authors' knowledge, there

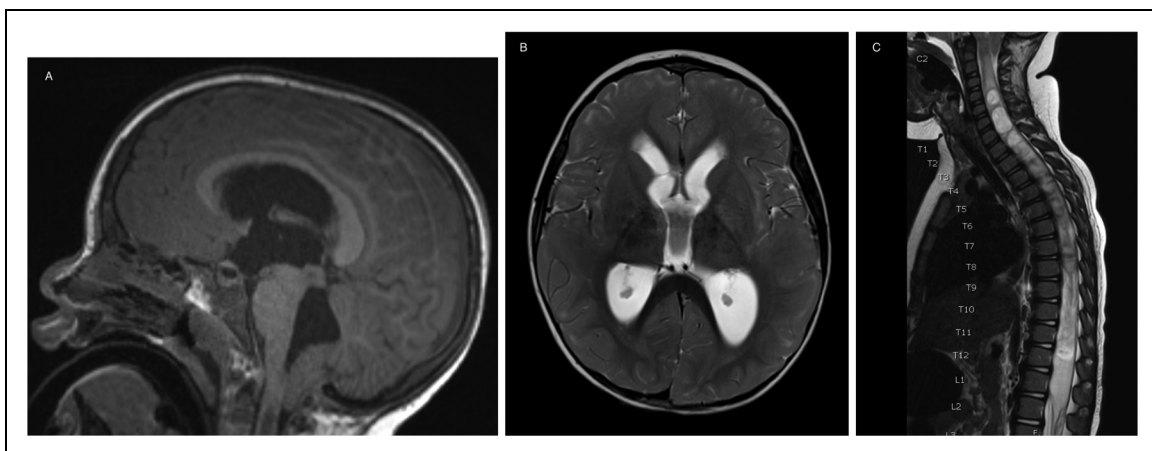


Figure 1. (A) Preoperative sagittal T1-weighted MRI of the brain shows Chiari I malformation (cerebellar tonsils displaced 13 mm below the foramen magnum) and hydrocephalus. (B) Preoperative axial T2-weighted MRI of the brain shows hydrocephalus. (C) Preoperative sagittal T2-weighted MRI of the C- and T-spine shows cord edema throughout the cervical spine, a syrinx from C3-4 extending inferiorly to T8-T9, and displaced peripheral parenchymal thinning extending to T8-T9.

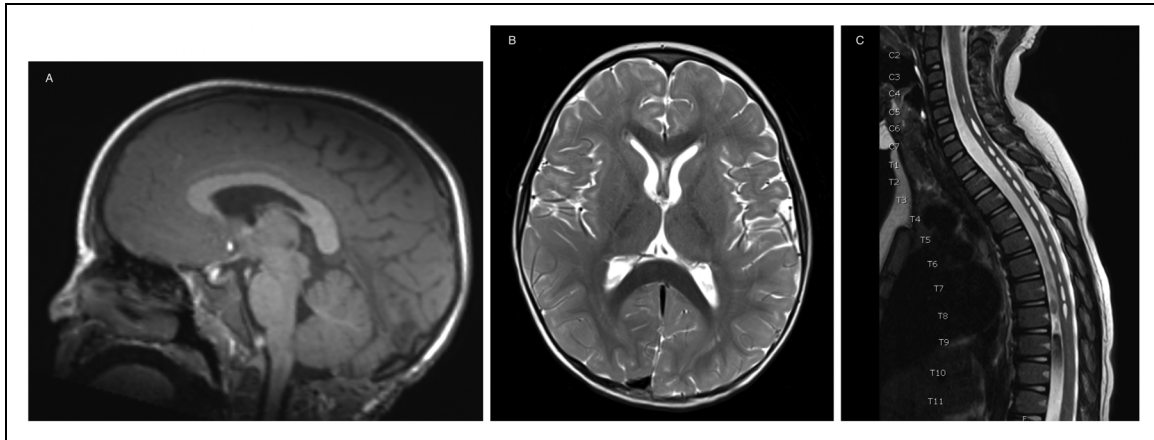


Figure 2. (A) sagittal T1-weighted MRI of the brain taken 2 months after surgery shows adequate hindbrain decompression and resolution of hydrocephalus. (B) Axial T2-weighted MRI of the brain taken 2 months after surgery shows resolution of hydrocephalus. (C) Sagittal T2-weighted MRI of the C- and T-spine taken 2 months after surgery demonstrate that holocord syrinx has decreased in size.

are no documented cases of Chiari I malformation with a concomitant large syringomyelia and severe hydrocephalus that presented with a clinical picture of spinal shock in a child under four years old.

In patients older than four years old, such presentations were rare with approximately nine reported cases globally.^{5–11} Similar to the present patient, the reported cases were found in patients with normal motor development. Patients presented with rapidly progressive leg weakness and loss of reflexes and were found to have holocord syrinx on MRI. However, these cases differed from the present case, as all previous cases were associated with unilateral foot drop. Bilateral presentation in this patient may be attributed to the size of the syrinx or its extensive expansion.

While rare in presentation, it is important to recognize, as misdiagnosis can delay proper treatment and lead to complications and poorer prognosis. Mitigating treatment delay is significant as syringomyelia prognosis after surgical decompression is dependent on deficits present prior to surgery. Delaying treatment is also associated with higher rates of postoperative complications and residual neurological deficits.¹²

It is important to recognize this clinical entity and differentiate it from other neurological disorders that can have a similar clinical presentation. In this case, the broad differential included Guillain-Barré syndrome (GBS), acute disseminated encephalomyelitis (ADEM), acute flaccid myelitis (AFM), central nervous system infection, acute cerebellar ataxia, spinal lesions, or neoplasm. A stepwise approach is recommended, with early acquisition of brain and spine MRI prior to pursuing more invasive procedures. Progressing with a lumbar puncture prior to imaging in this case may have exacerbated the herniation causing potentially severe complications.^{13,14} Studies have shown that reduction in CSF pressure in the spine may cause dramatic neurologic deterioration of individuals with Chiari I malformation.

Conclusion

This unique presentation of Chiari I malformation in a toddler underscores the importance of timely diagnosis, investigation

of differential diagnoses that can mimic this presentation, and misdiagnosis with delayed treatment.

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Ethics Approval

Our institution does not require ethical approval for reporting individual cases or case series.

Informed Consent

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ORCID iDs

Angelina Lo  <https://orcid.org/0000-0003-2040-2333>

Zurisadai Gonzalez-Castillo MD  <https://orcid.org/0000-0002-1800-2712>

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