Clinical Presentation of Chiari I Malformation and Syringomyelia in Children

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KEYWORDS

• Chiari I malformation • Syringomyelia • Syrinx • Scoliosis • Headache • Cervicomedullary junction • Craniocervical junction

KEY POINTS

- Chiari I malformation (CM1) may present with varying symptoms depending on patient age, or may represent an incidental finding in asymptomatic patients.
- The most common clinical presentations of CM1 in children involve occipital headache or posterior cervical pain, syringomyelia, and/or scoliosis.
- The constellation of symptoms and signs in CM1 relate to brainstem or cervical spinal cord compression, lower cranial nerve dysfunction, and/or syringomyelia.
- Clinical examination signs in children with CM1 may include sensorimotor deficits, lower cranial nerve deficits, or signs of medullary dysfunction.
- In rare circumstances, children with CM1 may present with acute onset or rapidly progressive findings warranting urgent surgical consideration.

INTRODUCTION

Chiari I malformation (CM1) refers to abnormal cerebellar tonsillar descent below the foramen magnum, typically at least 5 mm below this level for a qualifying diagnosis.[1–6] This form of hindbrain herniation may present with commonly recognized or rarely reported clinical symptoms, and also may be found incidentally. The frequency of radiographic findings consistent with CM1 among children undergoing brain and/or spine imaging for any indication has been estimated to be as high as 3% to 4%.[7] Symptoms from CM1 generally exhibit the following characteristics[7]:

- Clinical presentation (symptoms, signs) vary with respect to age; and
- Younger patients tend to present sooner, with shorter symptom duration, than adult patients.

As described more thoroughly in this article, tonsillar impaction within the foramen magnum causes compression of the cervicomedullary junction. The resulting compression and abnormal cerebrospinal fluid flow dynamics across the craniocervical junction result in a differential pressure gradient.[1–3,5,6,8,9] Thus, the clinical presentation of CM1 in children may reflect a constellation of symptoms resulting from compression at the cervicomedullary junction and those related to syringomyelia and scoliosis.[7,10,11] However, as with many neurosurgical conditions based partly on radiographic findings, CM1 may be discovered

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incidentally within asymptomatic patients or patients evaluated for nonspecific symptoms like headache or dizziness. Approximately 15% to 37% of pediatric patients with radiographic evidence of inferior cerebellar tonsillar ectopia may be asymptomatic, based on large retrospective reviews.[4,12,13] Additionally, patients may develop acquired tonsillar descent owing to other causes (posterior fossa mass, lumbar cerebrospinal fluid drainage/leakage, etc).

The most common clinical presentations of CM1 involve headaches and/or neck pain and scoliosis (most commonly levoscoliosis; **Table 1**).[2,4,7,8,12,14] Headaches represent a significant component of the clinical presentation in 27% to 70% of children with symptomatic CM1.[2,3,5,7,12,15] Classically, headaches in adolescents and adults demonstrate an occipitocervical location and are provoked or intensified by Valsalva-type maneuvers, with relatively ephemeral duration.[1,2,4–8,10,12,14] Various functions or activities that induce a Valsalva-type response, consistent with childhood behavior, include[7]:

- Sneezing, coughing;
- Laughing, screaming;
- Defecation; and
- Running, repetitive jumping.

Younger patients (neonates, infants, toddlers) often may fail to demonstrate or adequately communicate these classic headache descriptors.[12]

Characterized objectively by Cobb angles, scoliosis demonstrates strong association with syringomyelia in CM1.[6–9,14] In general, the following rules apply[2,8,9]:

- Most (but not all) pediatric CM1 patients with scoliosis have underlying syringomyelia; and
- Not all CM1 patients with syringomyelia have scoliosis.

Syringomyelia in CM1 demonstrates predilection for the following regions[2,3]:

- Cervical spinal cord (15%–21%),
- Cervicothoracic spinal cord (12%–25%);
- Thoracic spinal cord (15%–16%);

Table 1
Common clinical presentations of Chiari I malformation and syringomyelia in children

Clinical Sign or Symptom	Reported Prevalence in Pediatric CM1 Population[a]
Headache, neck pain	27%–70%
Scoliosis	18%–50%
Motor or sensory deficits of the extremities Paresis (typically in the upper extremities) Sensory deficits (in upper and/or lower extremities) Decrement of fine motor control	6%–17%
Irritability (typically younger patients)	12%
Oropharyngeal dysfunction, dysphagia, dysarthria Absent or reduced gag reflex Decreased palatal elevation Vocal cord dysfunction, hoarseness Tongue atrophy	4%–15%
Sleep apnea or bradycardia	4%–13%
Spasticity, hyperreflexia (typically in the lower extremities) Reflection of upper motor neuron impairment	6%
Frequent emesis	1%–10%
Ataxia or gait impairment, decreased coordination	4%–9%
Dysesthetic pain in C2 dermatomal distribution	7%
Facial numbness (trigeminal nerve sensory deficits)	7%
Auditory symptoms (tinnitus, hearing loss)	2%
Respiratory difficulty, dyspnea, stridor	1%–2%
Atrophy, hyporeflexia, and/or fasciculations in upper extremities Reflection of lower motor neuron impairment	Not reported

[a] Prevalence estimates are based on frequencies reported in peer-reviewed publications, and likely represent upper estimates in most cases.

Data from Refs.[2–4,7,8,10–12,14,15,17]