Chiari Malformation

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Introduction

The term Chiari malformation has accrued multiple meanings over time. Formally, this includes various groupings of hindbrain dysmorphism, with four numbered malformations (I–IV) which describe different cerebellar configurations. It is worth noting that while types I-III all include displacement of the cerebellar tonsils through the foramen magnum, type IV is cerebellar hypoplasia and likely a completely separate process from the other three—linked only by name. For the sake of simplicity: type I is displacement of the tonsils below the plane of the foramen magnum; type II is a far more complex malformation that includes displacement of the hindbrain, IVth ventricle, and cerebellar tonsils and vermis below the foramen magnum as well as a more diffuse constellation of brain anomalies with varying degrees of severity found in association with myelomeningocele; and, type III is similar to type II but found in association with a suboccipital or high cervical encephalocele. Patients with types II and III commonly have hydrocephalus. Historically, some have linked the work of Arnold with the type II malformation (Arnold-Chiari), although his contributions were minimal relative to Chiari. In addition, some groups have described a so-called Chiari 0, in which there are symptoms of brainstem compression or foramen magnum outlet obstruction without tonsillar herniation, presumably secondary to other anatomic structures such as arachnoid bands or scar [1].

Further note should be made of the terms *syringomyelia* and *hydromyelia,* which are often used interchangeably to describe the presence of fluid (presumably cerebrospinal fluid, CSF) within the substance of the spinal cord, a finding commonly associated with cases of Chiari malformation. Technically, if the fluid cavity is lined with ependyma (i.e., and expansion of the central canal), the term hydromyelia is used, while syringomyelia is defined as a fluid space within the spinal cord that is not lined by ependyma (also called a *syrinx*).

This chapter will focus on the Chiari I malformation (CM I), as it is the most commonly encountered type. There is a great deal of controversy surrounding the diagnosis and treatment of Chiari I, including the degree of displacement required for diagnosis (with most requiring at least 5 mm of herniation below the foramen magnum, but some accepting 0–2 mm) and what constitutes a pathologic Chiari I. Some experts have called for renaming the condition to Chiari "anomaly," as it is thought to be present in about 0.75 % of the population [1–3]. Here we will review pathologic Chiari I malformations with associated treatment and outcomes.

Key Points

Chiari I malformations are defined as displacement of the cerebellar tonsils at least 5 mm below the foramen magnum (Fig. 11.1).

The vast majority of Chiari I malformations are asymptomatic and do not need intervention [3].

When symptomatic, Chiari I malformations often present with sudden-onset suboccipital headaches classically aggravated by activities that invoke a valsalva maneuver. Less commonly, patients can present with lower cranial nerve dysfunction (especially dysphagia or sleep apnea), cerebellar dysfunction (e.g., ataxia), or spinal cord dysfunction secondary to an associated hydromyelia (weakness, scoliosis).

Treatment (when necessary) is predicated on surgical decompression of the cervicomedullary junction.

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R. Rahbar et al. (eds.), *Pediatric Head and Neck Tumors,*
DOI 10.1007/978-1-4614-8755-5_11, © Springer Science+Business Media New York 2014

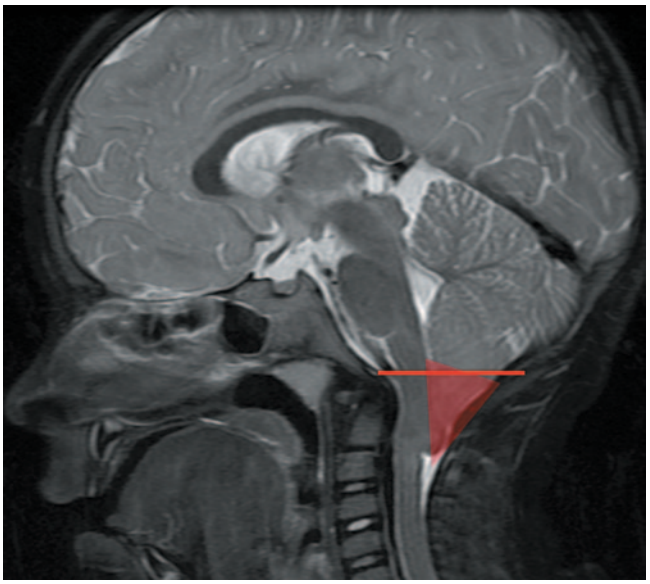

Fig. 11.1 Sagittal T2 magnetic resonance imaging (MRI) demonstrates presence of a CM I, with herniation of the cerebellar tonsils (*shaded in red*) below the foramen magnum (*red line*), compressing the cervicomedullary junction

Biology and Epidemiology

The etiology of CM I remains controversial. The majority of cases are considered to be developmental and a result of a mismatch between growth of the brain and that of the posterior fossa. Evidence comes from craniosynostosis patients and the observation of the clinical constellation of symptoms developing over time, as well as the observation that tonsillar displacement can change for better or worse over time during childhood, sometimes even resolving altogether [2, 4]. A minority of Chiari malformations can be acquired, presumably secondary to an abnormal pressure differential across the foramen magnum such as from hydrocephalus or a mass lesion from above or a lumboperitoneal shunt from below [5].

Pathophysiology

Symptoms from Chiari malformations are generally considered to be secondary to local compression or irritation. Tonsillar compression and scar-related tethering of the dura (particularly with valsalva maneuvers) irritate pain fibers in the dura, leading to headache [6].

Direct compression of the brainstem may contribute to dysfunction of local tracts and nucleui, leading to problems with swallowing, respiration, phonation, and other lower cranial nerve palsies.

Obstruction of normal CSF flow at the cervicomedullary junction has been implicated in the development of hydromyelia (or syringomyelia). This can cause chronic injury to pain and temperature fibers (which cross centrally in the spinal cord to produce a classic "suspended" sensory loss), compression of anterior horn motor neurons leading to lower motor neuron weakness (typically in the hands), compression of corticospinal tracts leading to upper motor neuron weakness with spasticity (typically in the legs), and possible scoliosis from weakness of the axial musculature [7].

Molecular/Genetic Pathology

- The majority of Chiari I malformations are thought to be sporadic, although 3 % of cases are found to be familial [8].
- There are no known genes specifically implicated in the cause of CM I, but links have been suggested to craniofacial disorders (including some craniosynostosis syndromes) and some connective tissue disorders [9, 10].
- Patients may have other findings, including Klippel-Feil, basilar invagination, or other areas of bony fusion [11].

Incidence and Prevalence

- Approximately 1/150 individuals will have some degree of tonsillar displacement potentially consistent with a CM I [1–3].
- Of all Chiari I malformations, only ~3 % are familial [8].

Age Distribution

- Mean age of presentation with symptoms = ~ 30 y/o
- Syrinx present in ~ 40 %
- Amount of tonsillar displacement correlated with symptoms:
  - Asymptomatic ~ 7 mm
  - Headache ~ 8 mm
  - Central cord symptoms ~ 10 mm
  - Brainstem compression symptoms ~ 12 mm [6, 12]

Sex Predilection

- There is a slight female preponderance at 3:2 [12].

Geographic Distribution

- None

Risk Factors—Environmental, Life Style

- None

Relationships to Other Disease States, Syndromes

- As noted above, there are several associations between Chiari I and other conditions [11]:
  - Hydrocephalus
  - Craniosynostosis
  - Endocrinopathies (growth hormone deficiency and acromegaly)
  - Hyperostosis
  - Bone mineral deficiency
  - Cutaneous disorders (neurofibromatosis type I, blue rubber bleb nevus)
  - Spinal defects (Klippel Feil, spondyloepiphyseal dysplasia)

Presentation

Symptoms/Signs

- Headache (occipital or suboccipital, tussive, worse with flexion/extension)—most common (about 2/3 of patients).
- Scoliosis was the 2nd most common reported finding in symptomatic Chiari I malformations in children [8].
- Lower cranial nerve dysfunction (hoarseness, dysphagia, dysphonia, aspiration, swallowing problems, snoring, apnea).
- Cerebellar syndrome (dysmetria, ataxia, nystagmus).
- Central cord syndrome (loss of pain and temperature sensation, weakness with lower motor nerve injury, scoliosis (often with syrinx)).
- Greater degrees of displacement often correlate with increasing severity of symptoms [6, 12].

Patterns of Evolution

- The presentation of symptoms in CM I is often chronic, over a period of months or years.

Evaluation at Presentation

- MRI is the current standard for evaluation of the cervicomedullary junction (CMJ).
- Discovery of a CM I often includes imaging of the brain to exclude mass lesions rostrally and—on occasion—MR imaging of the spine to assess for the presence and extent of syrinx.
- In some cases (such as those with a history of neck trauma), the history or imaging findings may suggest the need to assess the bony anatomy of the CMJ with other diagnostic studies, such as flexion–extension radiographs or computerized tomography.
- Standard preoperative laboratory studies (complete blood count (CBC), clotting times Prothrombin Time/Partial Thromboplastin Time (PT/PTT), type and cross (T&C) for blood bank, chemistry panel (Chem 7)) may be considered prior to planned surgery.

Differential Diagnosis

Many patients will be asymptomatic on examination if the lesion is found incidentally. The diagnosis of a CM I is relatively straightforward, with the difficulty primarily arising from the exercise of using clinical judgment to select appropriate surgical candidates. The main issue with differential diagnosis is to ascertain whether a proximate cause for the CM I exists. As such, imaging of the head and spine may be warranted to exclude intracranial mass lesions, hydrocephalus or areas of spinal CSF leak. Furthermore, careful history taking is important to determine if things such as lumbar puncture or intracranial pressure (ICP)-elevating medications (such as retinoic acid) may be contributing to the radiographic findings. Lastly, given the often subjective nature of complaints in CM I, the clinician must carefully evaluate the patient for other causes that may explain presenting symptoms.

Diagnosis and Evaluation

Physical Examination

- Many patients will be asymptomatic on examination. However, a detailed neurologic examination and history are always important. Attention should be paid to evidence of neurologic dysfunction in the history.
  - Headache—usually occipital and tussive which can often be replicated on exam, need to assess whether the pain is related to flexion or extension of the neck
  - Lower cranial nerve dysfunction—apnea (infants), snoring, dysphonia, dysphagia, trapezius weakness
  - Spinal cord dysfunction—weakness in the arms or legs (especially the hands with signs of muscle wasting), long tract signs (with hyperreflexia Babinski, “cape” sensory loss distribution), scoliosis

Laboratory Data

- Standard preoperative laboratory studies (complete blood count (CBC), clotting times (PT/PTT), type and cross (T&C) for blood bank, chemistry panel (Chem 7)).

Imaging Evaluation

MRI is the imaging modality of choice in the evaluation of CM I. If possible, studies of the brain should be obtained (either MRI or computed tomography, CT) to exclude the possibility of mass lesions or hydrocephalus as proximate, treatable causes of CM I. The use of high-resolution MRI (such as Fast Imaging Employing Steady State Acquisition (FIESTA)) may be useful in assessing for the possibility of obstruction to the outflow of the 4th ventricle. Contrast is usually not needed, although it may be helpful in assessing the location of the confluence of sinuses in the posterior fossa or the choroid in the 4th ventricle (as well as excluding other lesions such as tumor or vascular anomalies, if suspected).

In addition to the objective measurement of tonsillar herniation, there are subjective measures that can be assessed using MRI to help determine the extent of compression. These subjective findings include the presence of "peg-like" tonsils, obliteration of CSF spaces at the CMJ, the presence of a syrinx (sometimes requiring dedicated spinal imaging) and the quantification of CSF flow.

CSF flow studies have been considered as diagnostic tools when considering surgical decompression of Chiari malformation. In patients who clearly have symptoms related to their Chiari, diminished CSF flow in preoperative assessment appears to correlate well with surgical outcome. There is no good evidence, however, that CSF flow studies have any advantage over clinical impression in determining whether a Chiari malformation is symptomatic [13].

CT and plain films are generally not warranted unless there is a concern for basilar invagination, atlanto-axial instability, or other bony injury that might not be readily assessed with MRI. However, in some cases, such as reoperations (in which the extent of bony decompression may need to be seen) or in situations that might require arthrodesis (such as those children with rheumatoid arthritis), CT or flexion–extension plain films can be helpful.

Nuclear medicine tests

- Not usually warranted with CM I

Electrodiagnostic tests

- Not usually warranted with CM I

Neuropsychological tests

- Not usually indicated with CM I

Treatment

Goal To remove compression at the cervicomedullary junction, restore physiologic pulsatile CSF flow, and thereby alleviate symptoms and/or provide protection from future injury.

Surgical therapy

Surgical Indications One of the greatest challenges in CM I is appropriate selection of surgical candidates. "The Chiari malformation is, in fact, one of the few conditions for which the AANS (American Association of Neurological Surgeons) issued a position statement regarding the inappropriate use of surgery (AANS Position Statement on the Use of Cervical Decompression for Chronic Fatigue Syndrome, March 2000). When a patient presents with two common conditions, there is always going to be some degree of coincidental overlap, and surgeons must be careful not to perform surgery in patients in whom there is little chance that the Chiari malformation is symptomatic [2]."

In general, operation is indicated in patients with associated symptoms (see above) and clear radiographic evidence of disease (>5 mm herniation, +/− syrinx). For those with atypical symptoms (frontal headache, fatigue, etc.) and minimal radiographic findings, a conservative approach with a referral to pain management might be appropriate. Surgery for asymptomatic patients (including incidentally found lesions) remains controversial and has been justified on the basis that this lesion has the potential to become symptomatic or could place the patient at greater risk of spinal cord injury if left untreated, although this has not been substantiated.

One of the factors to support surgical intervention for asymptomatic Chiari includes prevention of an exacerbation after trauma. Development of symptoms related to a Chiari after a minor traumatic event has occurred, with reports indicating approximately 13 % of previously asymptomatic CM I developing symptoms after trauma (including isolated case reports of sudden death in severe cases) [14, 15].

Athletes with Chiari malformation present additional challenges in the determination of appropriate management for an incidentally found Chiari. Numerous athletes have been found to have Chiari malformation on imaging obtained after suffering a concussion, and reports of athletes with known Chiari experiencing drop attacks have been reported. The implication of these associations is unclear, and it has yet to be determined whether athletes with asymptomatic Chiari who play contact sports are at a greater risk for catastrophic injuries. A known Chiari with clear associated symptoms is often considered a contraindication to contact sports. Asymptomatic Chiari malformation may be a rela-

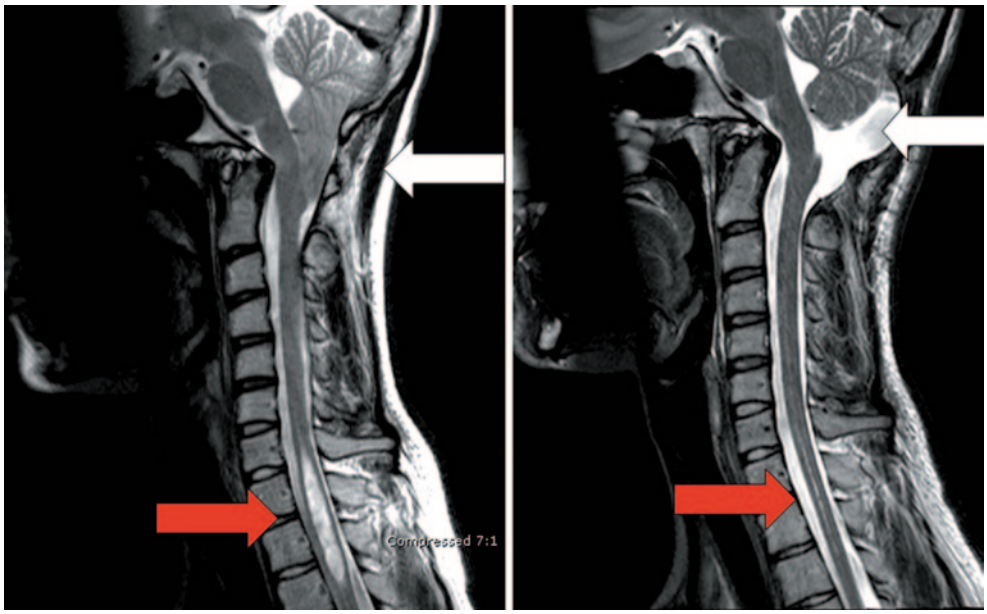

Fig. 11.2 Sagittal T2 MRI of the posterior fossa and cervical spine showing preoperative images (*left*) with an extensive CM I (*white arrow*) and cervical cord syrinx (*red arrow*), coupled with postoperative images from the same patient 6 months later (*right*), revealing decompression of the cervicomedullary junction (*white arrow*) and marked reduction in the syrinx (*red arrow*)

tive contraindication due to increased risk of injury based on anecdotal evidence. A careful discussion with the athlete and family is important. Most athletes can return to sports after undergoing a surgical Chiari decompression.

Surgical Techniques

The method of decompression is also a topic of debate. Generally, a suboccipital craniectomy is performed, often with removal of the posterior ring of C1. Controversy exists as to the utility of opening the dura and placing a dural patch, with some authors calling for the use of intraoperative ultrasound to assess the extent of decompression after bony removal and using the findings to guide the need for dural opening. Overall, 7% of pediatric neurosurgeons only perform bony decompression (without dural opening), while 36% always open the dura [16].

Relieve external pressure on the crowded foramen magnum region
- Suboccipital craniectomy, C1 laminectomy
- Open dura and place dural graft (either routinely or only after ultrasound)

Reestablish normal CSF flow out of IVth ventricle in patients with syringomyelia
- Remove scarring from IVth ventricle outflow
- Possible "shunt" from IVth ventricle to cervical subarachnoid space

Complications [16]

Overall, published complication rates for pediatric CM I operation range from 2–40% [8, 17].

- Bleeding is the most immediate complication of surgery and risks are magnified in smaller children, who have little reserve. In particular, high venous pressures and large dural sinuses in the posterior fossa increase the risk of hemorrhage.
- CSF leak can occur when dura is opened, including pseudomeningocoele.
- Chemical meningitis.
- Vertebral artery injury.
- "Cerebellar slump" is a controversial entity often ascribed to excessive decompression of the posterior fossa, resulting in sag of the tonsils and recrudescence of Chiari-related symptoms.

Outcomes

Outcome After Surgery (Fig. 11.2)

- In a recent large series, average hospital stay and "return to school" time after surgery were 3 and 12 days, respectively [8].
- In carefully selected, symptomatic patients, 60% will demonstrate durable improvement in symptoms following surgery [18].
- In patients with symptomatic syrinx, over half will experience improvement clinically within 10 months [19].
- Three percent of patients will need reoperation for recurrence (more likely with younger patients) [8].

Follow-up

Frequency of Office Visits

- Postoperative care will frequently consist of an office visit approximately one month postoperatively.
- Rare long-term issues include recurrence and craniocervical instability. While most patients can be discharged from care after routine postoperative visits, some may require long-term follow-up.

Conclusions

The management of CM I remains controversial, with debate centered on operative indications and surgical approach. Candid discussions with patients and families are useful in establishing realistic plans of action and expected outcomes.

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