The Chiari Malformation: What Does It Mean?

***(also, see PowerPoint presentation* "The Chiari Malformation: Incidental Finding or Pathological Process?")**

Joseph H Piatt, Jr, MD, FAAP
Chief, Section of Neurosurgery
St Christopher's Hospital for Children
and
Professor of Neurological Surgery and Pediatrics
Drexel University College of Medicine

Introduction

The Chiari malformations were first described in 1890 by the German pathologist whose name they bear, and their relationship to progressive neurological disability has been understood almost as long. The condition was recognized infrequently, however, because the radiological tests required were difficult both for the radiologist to conduct and for the patient to endure. Only the most desperately ill ever received a diagnosis.

What has changed in the last 20 years is the development and dissemination of magnetic resonance (MR) imaging technology. MR imaging yields detailed pictures of any part of the body without risk or discomfort (although young children and some claustrophobic adults may require sedation) in sessions that generally last less than an hour. The enormous diagnostic power of MR imaging has blossomed, in this country at least, in a very fertile economic environment for medical imaging services, and MR imaging units are now everywhere. The clinical threshold for ordering MR imaging has fallen to a very low level, and patients with mild symptoms and with no neurological impairment are routinely subjected to studies of the brain and spine.

MR imaging has demonstrated that the Chiari malformations are rather common among patients who have undergone diagnostic imaging for unrelated reasons. The prevalence in the general population has been estimated at a little less than 1:1000. Not surprisingly, since the Chiari malformation seems to be present in so many people who are otherwise perfectly well, MR imaging has also opened the possibility of associations between the Chiari malformation and a long list of nonspecific and nondisabling neurological symptoms. In the modern era the challenge is not the diagnosis of the Chiari malformation but the selection of patients who really require treatment from among the many who carry the diagnosis.

Hans Chiari actually described 4 types of brain malformation. The 3rd and 4th types are medical curiosities only. The 2nd type is found exclusively in association with myelomeningocele, the most severe form of spina bifida. Its management is inextricably commingled with management issues arising from the other complications of this condition, and it is beyond the scope of this article. The Chiari malformation type 1 (CM1) is the entity whose recognition and treatment have been so transformed by the technology of MR imaging, and what follows is a review for patients and parents of the anatomy, physiology, natural history, and management of this condition.

Anatomy and Physiology

The brain belongs inside the skull. The various parts of the brain reside in their corresponding compartments within the cranial cavity, and brain and the cranium normally grow hand-in-hand and fit each other perfectly. In the CM1, however, the cerebellum and the brainstem, portions of the brain sometimes referred to collectively as "the hindbrain," do not fit inside their proper compartment, the posterior cranial fossa (**Figure 1**). As they grow, the cerebellum to a lesser degree, the brainstem are squeezed out of the skull through the foramen magnum, the large hole at the base of the skull through which the spinal cord passes to its junction with the brain stem. This protrusion of the cerebellum is vividly demonstrated by sagittal MR imaging of the head or the neck, and displacement of the cerebellum more than 5mm below the plane of

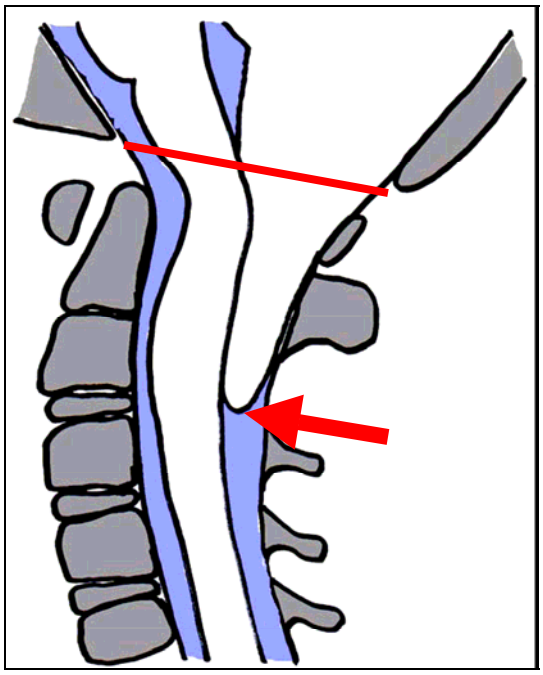

Figure 1. *In this cartoon of the Chiari malformation type 1 (CM1), the bones of the skull and the spine are gray, the brain is white, and the cerebrospinal fluid is blue. The red bar marks the level of the foramen magnum, the hole at the base of the skull through which the spinal cord passes. The red arrow marks the tips of the tonsils of the cerebellum, the portion of the brain that is displaced in the CM1.*

the foramen magnum is considered abnormal. The 5mm limit is the customary definition of the CM1.

As this definition suggests, some Chairi malformations are worse than others. In some instances the inferior portions of the cerebellum, also called the "tonsils" of the cerebellum, can protrude several centimeters into the cervical spinal canal. In such severe cases the tonsils are compressed and deformed, resembling a cork squeezed into the neck of a wine bottle. As the cork analogy implies, the displaced cerebellum and the adjacent brainstem and spinal cord may be severely compressed. Impairment of the function of these neural structures may cause symptoms and disabilities, and signs of tissue damage may be visible on MR imaging.

Probably the most important complication of the CM1 is syringomyelia (**Figure 2**). Syringomyelia is cavitation of the spinal cord. "Syrinx" is the term for an individual cavity. The CM1 is the result of a developmental aberration, so in most instances it is present at birth. Syringomyelia, on the other hand, is acquired later in life, and to understand how the CM1 causes the appearance of syringomyelia requires an explanation of the physiology of cerebrospinal fluid (CSF). CSF is the salt-water solution in which the brain and spinal cord float. It is contained by a thin, tough membrane, called the "dura," that lines the spinal canal and the inner aspect of the skull. The brain produces CSF within internal cavities, called "ventricles," and after it has flowed out of the ventricular system and has bathed the brain and spinal cord, CSF is recycled into the venous side of the circulation. CSF is in constant motion. Not only is it flowing slowly from the site of its secretion to the site of its recycling, but it also pulsates in a to-and-fro manner with each heart beat.

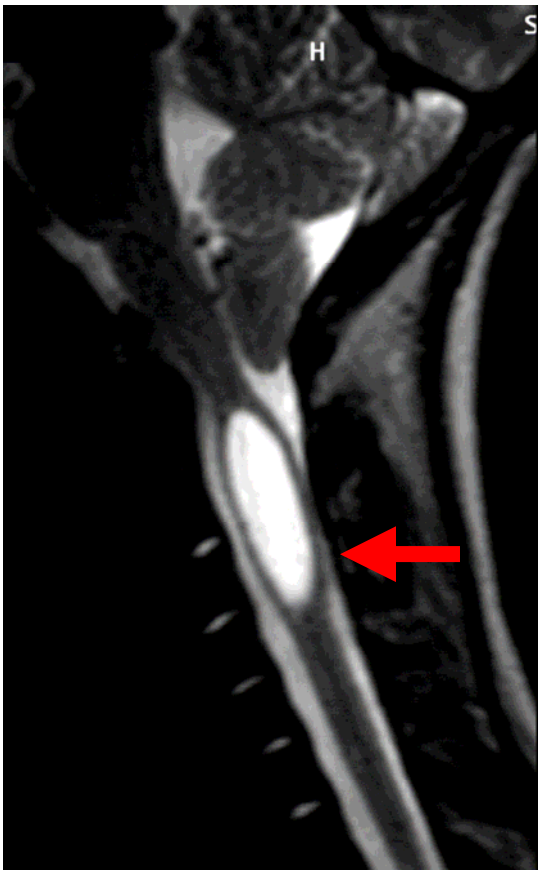

Figure 2. *This sagittal MR imaging view of the CM1 shows the same portion of the skull and cervical spine as Figure 1. The CSF is light gray or white in this imaging sequence. The red arrow marks a distended syrinx cavity that widens the silhouette of the spinal cord.*

Altogether the progress of CSF resembles a conga line. The brain itself pulsates with each heart beat as well, and if the tonsils of the cerebellum are jammed snugly into the upper cervical spinal canal, they act like a piston. With each heart beat the downward stroke of the cerebellar piston causes an abnormal surge in the pressure of the CSF within the spinal canal. Syringomyelia develops as CSF is driven into the substance of the spinal cord by this extreme pulse pressure. The CSF collects inside syrinx cavities that expand gradually by stretching and tearing the surrounding spinal cord tissue. MR imaging is very sensitive in detection and demonstration of syringomyelia.

In most instances no underlying cause for the development of the CM1 ever comes to light, but in some patients the CM1 appears as a feature of another recognizable syndrome or condition. Table 1 is a list of the most common. They fall into 2 general categories: conditions that affect skull growth and conditions that disturb CSF circulation. Crouzon and Pfeiffer syndromes are the most common of a class of genetically determined disorders of the development of the craniofacial skeleton. They are associated with obvious deformities of the face and head. In the usual sequence of events, the CM1 is disclosed by investigations set in motion because of the syndromic diagnosis, not the other way around. Likewise, recognition of achondroplasia and other skeletal dysplasias usually precedes the discovery of an associated CM1. The effect of rickets, the bone disease caused by vitamin D deficiency, on the growth of the skull can be more subtle. Skull deformity in this condition may be inconspicuous or absent altogether, in which case the diagnosis has usually been made on the basis of deformity of other portions of the skeleton or detection of low blood calcium levels. In rare instances the diagnosis is only inferred: Skull disease and associated CM1 due to rickets is particularly common in children of North African extraction, for instance, probably due to a combination of racial and dietary factors. Hydrocephalus, a class of conditions characterized by disturbed CSF recycling, is present in 10 to 15% of patients with CM1 and must be treated in conjunction with it. Rarely the CM1 is associated with conditions that cause low intracranial pressure. In such instances the cerebellum is not pushed out of the skull but sucked out of the skull because of abnormal leakage or drainage of CSF from the spinal canal below. The hallmark of low intracranial pressure is a severe headache precipitated by assuming an upright posture and relieved by recumbency. Finally, about 10% of patients with CM1 have a first degree relative with CM1 without any other recognized genetic or inherited condition.

Table 3 – Conditions associated with CM1

Conditions affecting skull growth
- Crouzon and Pfeiffer syndromes (and other craniofacial dysostoses)
- achondroplasia (and other skeletal dysplasias)
- rickets

Conditions disturbing CSF circulation
- hydrocephalus
- intracranial hypotension
  - spontaneous
  - lumboperitoneal CSF shunting

Symptoms and Disabilities

The CM1 can cause symptoms either through compression of neural structures by the displaced cerebellar tonsils or through damage to spinal cord tissue by syringomyelia. The possibilities are legion. Table 2 lists symptoms that can be caused by, or have been attributed to, tonsillar compression.

Table 2 – Symptoms of cerebellar tonsillar compression

headache
neck pain
inconsolable crying
torticollis or other less severe limitation of neck motion
difficulty swallowing
difficulty feeding
difficulty speaking
irregular breathing
weakness of eye movements, especially downgaze
incoordination of limb or trunk movements

Table 3 lists symptoms that can be caused by syringomyelia.

Table 3 – Symptoms of syringomyelia

neck or back pain
abnormal sensation in limbs or trunk
pain in limb or trunk
loss of sensation in limbs or trunk
limb weakness
muscle wasting
gait disturbance
incoordination of limb or trunk movements

spinal curvature (scoliosis) incontinence or other disturbance of bladder function disturbance of sexual function

These lists are long, and a moment's reflection will show that most of the symptoms on the lists are nonspecific in the sense that they have many other, more frequent, explanations.

The question of headache deserves special attention. Almost everyone has headaches at some time or another. The 2 most common headache patterns are muscular (or "tension") headaches and vascular (or "migraine") headaches. Tension headaches are experienced typically on the sides of the head or at the back of the head where the neck muscles attach to the skull. In some people they tend to be triggered by stress. They can be relieved momentarily by application of pressure at the painful site, and they usually respond to over-the-counter, nonnarcotic analgesics such as acetaminophen (Tylenol® and other brands), acetyl salicylic acid (Aspirin® and other brands – not for children!), and ibuprofen (Motrin®, Advil® and other brands). Migraine typically affects one side of the head. Patients often describe their headache as "pounding" in quality. It is typically associated with nausea, vomiting, and sensitivity to bright lights and loud noises. Less commonly but more specifically the headache may be preceded or accompanied by neurological symptoms, such as the perception of bright lights or black spots, or by abnormal sensations or weakness on the side of the body opposite the headache. Patients suffering a bad migraine often withdraw to a dark room and try to sleep, and in many cases sleep relieves the symptoms. Most patients with migraine have first degree relatives with headaches as well. The over-the-counter medications that are effective for tension headaches are often useful in treating mild, infrequent migraines, but for more severe and frequent migraines there is a class of specific, prescription medications that can prevent or interrupt the headache. Response of symptoms to one of these specific migraine medications can be considered a confirmation of the diagnosis.

The point of this digression into the phenomenology of headaches is that labels such as "tension headache" and "migraine" are only generalizations with indistinct boundaries. The headache characteristics that are the most definitive are also the least common, and many patients with chronic headaches do not fit neatly into one category or the other. Likewise for headaches attributed to CM1. The textbook description is a headache at the nape of the neck occurring in brief paroxysms precipitated by coughing, laughter, vigorous physical exercise, or straining to defecate. The mechanism for the production of pain is hypothesized to be abnormally large and abnormally sustained pressure differences between the cranial cavity and the spinal canal caused by the plugging of the craniospinal junction by the displaced cerebellar tonsils. In fact, in contemporary practice, very few patients with CM1 describe their headaches this way. How did this concept of the typical CM1 headache arise? It arose in the pre-MR imaging era when only the sickest patients or the patients with the most suggestive symptoms underwent the difficult and painful diagnostic tests needed to make the diagnosis. In the era of accessible MR imaging, the spectrum of symptoms that patients report is much broader.

The reader must now acknowledge the subtle challenge entailed in the distinction between association and causation. Headaches are common, and as MR imaging has revealed, the CM1 is fairly common too. Furthermore, headaches are a very common reason for patients to undergo MR imaging. How then can a doctor tell whether any particular kind of headache might be caused by the CM1? There is no scientific answer to this question. A clinical researcher might use rigorous statistical methods to compare the words that a large group of patients with the CM1 use to describe their headaches with the descriptors utilized by an equally large group of patients with normal MR imaging studies, and perhaps qualitative differences might be discovered. But this investigation has never been conducted. Even in the case of the headache that disappears after surgical treatment, the skeptic may legitimately suggest that the operation disrupted some other headache-generating mechanism involving the anatomic structures that were subjected to surgical manipulation – the skin, the muscles and ligaments of the neck, the dura, and the nerves in the neighborhood of the craniocervical junction – and that the CM1 was merely a coincidental finding. Clearly, clinical judgment must be exercised in the attribution of headache – or any other symptom – to the CM1, and some degree of uncertainty will always remain.

Natural History

The course that a condition follows over time without treatment is referred to as its "natural history." For patients who have intolerable symptoms or severe disability at the time of diagnosis, the natural history of the CM1 is not a great issue. They need treatment without delay. For the many patients who are discovered to have the CM1 after MR imaging for nonspecific symptoms such as headache or in the course of an evaluation of a related condition, such as a craniofacial dysostosis, the natural history is arguably the only important issue. Unfortunately, this matter has been studied inadequately, and the only available information is anecdotal in character and is further limited by the relatively recent proliferation of MR imaging units.

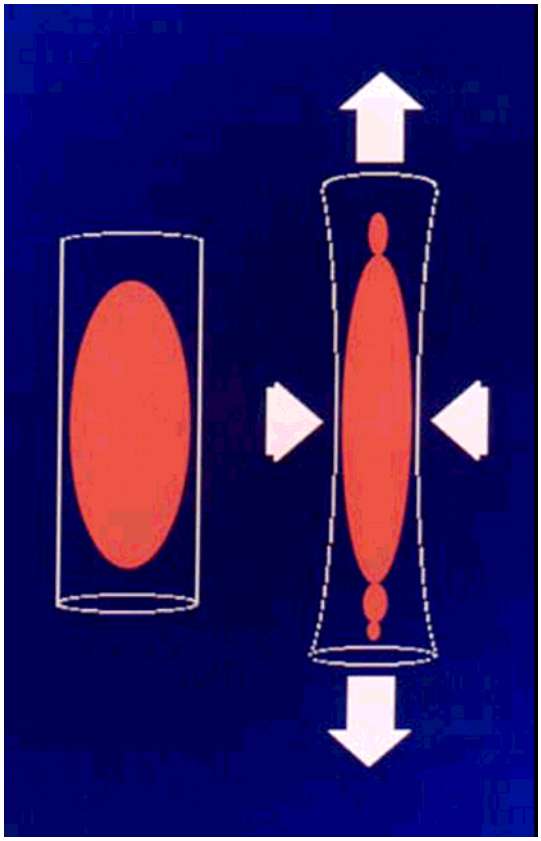

Figure 3. *Weight-lifting has been reported to cause rupture of syrinx cavities into adjacent segments of the spinal cord. Straining to life a heavy object raises pressure within the abdomen. Abdominal pressure is transmitted to the network of veins that surround the dural sac within the spinal canal. Distention of these epidural veins squeezes the spinal cord.*

A few general comments: The severity of the CM1 tends to remain stable over time periods as long as a few years. There are case reports of individual patients whose cerebellar displacement has either progressed or receded for no obvious reason, but such stories are rare. Of greater concern is the *de novo* development of syringomyelia in the CM1 patient without spinal cord involvement at initial diagnosis. This complication is a realistic concern, but how often and over what time period it can occur are not known. One might suspect that the risk of syringomyelia is related to the severity of the CM1 as measured either by the degree of cerebellar displacement or by the severity of the disturbance of CSF flow, which can be assessed by special MR imaging techniques. Neither of these hypotheses has been tested adequately. Finally, because the CM1 may involve some degree of persisting spinal cord compression by the displaced tonsils of the cerebellum, concerns have been expressed about the vulnerability of CM1 patients to catastrophic spinal cord injury from what would otherwise be considered minor head or neck trauma. There are a handful of reports in the medical literature of sudden death (presumed due to ventilatory arrest) or paralysis after rear-end automobile accidents or minor athletic injuries in patients who are subsequently discovered to have the CM1. Do these reports represent coincidences? Probably not, but they are very, very rare. The weight that they deserve in decisions regarding treatment and restriction of activities in untreated patients is not at all clear.

The natural history of syringomyelia is better understood, at least in the more severe cases, from experience accumulated in the pre-MR imaging era. Patients who have symptoms, neurological abnormalities, or functional disabilities tend to get worse slowly over months and years. Deterioration is typically step-wise, with long periods of stability interrupted by the sudden appearance of new problems. In some instances these sudden events seem to be precipitated by straining, as though abdominal pressure is transmitted to the spinal canal and squeezing of the syrinx cavity causes it to rupture into previously uninvolved segments of the spinal cord. For this reason patients with significant syrinx cavities are often counseled

not to lift weights or wrestle (**Figure 3**). What is not so well understood is the natural history of mild cases. Thin syrinx cavities that do not distend the spinal cord often went undiscovered before MR imaging, but with the aid of this technology they seem rather common and are usually asymptomatic. This writer's impression is that such patients and their thin syrinx cavities remain stable over periods of a few years, but their prognosis cannot yet be projected over a longer time frame.

Management

As the preceding discussion makes plain, there are important gaps in our understanding of the CM1, but decisions about management of this condition must be made nevertheless. When knowledge is incomplete, physicians must feel their way based on reasoning and experience, and variation of practice from physician to physician is great. What follows is a description of this writer's perspective on the most important management questions. The reader must understand that at almost every point a contradictory opinion might be found either in the published medical literature or at the offices of other neurosurgical practitioners. Hopefully, as medical knowledge advances, this document will require revision soon to reflect growing consensus about the most critical matters.

Because of the association of the CM1 with both hydrocephalus and syringomyelia, every patient deserves complete imaging of the brain and spine before treatment decisions can be addressed. About 10% of patients with CM1 have associated hydrocephalus, and treatment of the CM1 must not be undertaken without preceding or simultaneous treatment of the hydrocephalus.

When the relationship of symptoms to the CM1 is clear, and when symptoms are intolerable, the need for treatment is indisputable. If symptoms are mild, and, in this writer's view, if there is no syringomyelia, a decision about treatment may be deferred safely in favor of clinical and imaging observation. If the relationship of symptoms to the CM1 is not certain, judgment is required. Most often the symptom that creates such a degree of uncertainty is headache. This writer's usual practice is to encourage a vigorous attempt at pharmacological control of symptoms under the supervision of a child neurologist. For medically intractable headaches that are disabling, that is, headaches that interfere with school or other important activities, surgical treatment may be considered.

The child with syringomyelia at the time of diagnosis deserves special consideration. There is little disagreement that surgical treatment is indicated for the child with neurological disabilities, scoliosis, or (rarely) neuropathic pain. The question of asymptomatic syringomyelia is more problematic. Some surgeons withhold treatment unless symptoms develop or unless there is imaging evidence of expansion of the syrinx cavity. This writer usually recommends treatment of the CM1 even for asymptomatic syringomyelia. This writer views syringomyelia as a destructive and progressive process. Although long periods of clinical stability are common, eventual neurological losses are the expectation, and such losses are not necessarily reversible by subsequent treatment. As shall be seen below, surgery is effective and very safe. In the balance proactive intervention seems reasonable.

If treatment is deferred, the patient deserves surveillance for the development of new symptoms and disabilities and for the development or progression of syringomyelia. The schedule for follow-up is a matter of judgment based on the severity of the CM1 and the age of the patient. MR imaging of the cervical spine is the appropriate surveillance study for the development of syringomyelia. Surveillance imaging of the head is not necessary. Imaging surveillance does not need to be so frequent as office visits, and the interval between encounters can be lengthened gradually if the patient remains stable. As has been noted, the natural history of the CM1 is not known on long time scales. Whether a patient can ever be discharged from neurosurgical care is uncertain, but the writer's practice is to continue follow-up of the stable patient into late adolescence.

A vexing question is whether the child with CM1 should be subjected to restrictions of activity. The concern about sudden death or catastrophic paralysis after minor injury has been discussed above. Such events are exceedingly infrequent. The surgeon must exercise some judgment based on the severity of the CM1 on MR imaging and on the presence or absence of associated skeletal anomalies at the craniocervical junction. The nature of the activity must be weighed as well. The inclination of most pediatric neurosurgeons is to encourage vigorous physical activity

including participation in athletics, but collision sports may be unwise for the child with a severe malformation. Directors of high school and college athletic programs may sometimes request written assurance that the child with CM1 is at no greater risk of injury than any other child. What they are really asking is that the physician's malpractice carrier indemnify their athletic program against claims arising from Chiari-related injuries. This writer has not always been able to comply with such requests. Syringomyelia associated with the CM1 is a special case: Patients should refrain from weight-lifting and wrestling because of theoretical concerns, reinforced by anecdotal reports, about rupture and extension of the syrinx cavity precipitated by straining.

Surgery

The goal of surgery is to relieve compression of the brainstem and the spinal cord by the displaced cerebellum and to restore CSF pulsation back and forth across the craniocervical junction. The details of surgical technique vary greatly from surgeon to surgeon, and no 2 techniques have ever been compared in a scientific fashion. This writer begins with a skin incision in the midline of the back of the neck beginning at the top border of the neck muscles and extending midway down. The muscles are separated from each other in the midline and peeled off the base of the skull and the back of the first cervical vertebra. Bone is removed from the base of the skull to enlarge the foramen magnum, which is the opening through which the spinal cord enters the cranial cavity. A piece of bone about the size of a silver dollar is removed. The back portion of the first cervical vertebra is removed as well. Some surgeons believe that the operation can be concluded at this point, but the tonsils of the cerebellum are still down in the cervical spinal canal, compressed and deformed by the dura, which does not necessarily relax after the overlying bone has been cut away. The dura is now opened to expose and decompress the displaced cerebellar tissue. With the assistance of the operating microscope, this writer coagulates the cerebellar tonsils with electrical current that causes the surface of the tonsils to shrink. The tonsils become rounded, instead of flattened as they had been, and they retract into the skull cavity. The spinal cord and the brainstem are now well decompressed, and there is ample space for CSF to flow. Between the tonsils is an important outlet for CSF flowing out of the ventricular system of the brain, and in some cases of the CM1 this outlet is obstructed by an abnormal membrane. Continuing with the microscope this writer examines the ventricular outlet and relieves any obstruction that may be present. To maintain the decompression that was achieved by opening the dura, the dura is now reconstructed with a patch. A variety of natural and artificial materials have been used for patching the dura, but this writer currently favors a thin sheet of Gortex®. The bone is not replaced. It is not needed for protection of the exposed portion of the brain, which lies under a thick layer of neck muscle. The wound is finally closed in anatomic layers with absorbable suture material. The only dressing is a layer of antibiotic ointment. No hair has been shaved, and in fact the hair may be washed any time after the first postoperative day. Patients usually remain in the hospital for 3 to 5 nights – more often 3 than 4 or 5.

Fortunately the risks of surgical treatment of the CM1 are small. As with any operation there are very small risks related to general anesthesia. Blood transfusion is very seldom necessary, even for very young patients. CSF may collect under the skin incision after surgery in a puddle referred to as a "pseudomeningocele." Pseudomeningoceles usually heal without specific treatment, but in less than 5% of cases a return trip to the operating room for repair of the wound closure is necessary. There is a risk of wound infection, probably less than 1%, most often related to leakage of CSF out the wound. Wound infection is a very serious concern because it can spread to the CSF and the brain; it can be damaging to the brain or even life-threatening. The risk of direct, surgical injury to the brainstem or spinal cord is very, very small, but such injuries may be catastrophic. Paralysis, inability to speak, to swallow, or to breath without ventilator support are all theoretical possibilities. There are no meaningful statistics about the frequency of such severe neurological disabilities or death after surgery for the CM1, but the risk must be under 1 in 1000.

How effective is the surgical treatment of the CM1? Decompressive surgery relieves symptoms and signs caused by brainstem and spinal cord compression very reliably, but such a statement begs the question whether the CM1 is really the cause of the individual patient's complaints. Because so many patients receive

the diagnosis of CM1 after MR imaging obtained for nonspecific symptoms, there is often a degree of uncertainty about what benefit may be expected. The surgeon, the parents, and the patient, if possible, must all exercise some judgment. Happily, the results of treatment for syringomyelia complicating the CM1 are more objective, at least from an imaging standpoint, and they are very good. Decompressive surgery as described above leads to collapse or disappearance of syrinx cavities on MR imaging in about 80% of cases (**Figure 4**). As a general rule a visible decrease in the volume of the syrinx cavity on follow-up imaging means that the progression of the disease has been arrested. Relief of symptoms, disappearance of neurological abnormalities, and correction of spinal curvature are difficult to predict. Mild problems of short duration are more likely to remit than severe, long-standing problems. Rarely, symptoms and disabilities may continue to progress despite what appears from the standpoint of MR imaging to be successful treatment; the basis for continuing disease activity in this situation is not understood.

A small minority of patients with CM1 have persisting symptoms after surgery because of elevated intracranial pressure, the *pseudotumor cerebri* syndrome. (The term *pseudotumor* reflects that fact that the patient behaves as though she has a brain tumor, but there is no tumor.) In addition to headaches there may be visual symptoms, and physical examination may show swelling of the optic nerves visible at the back of the eyes. Typically there is no abnormality on brain imaging, and there is no abnormality in the CSF on laboratory examination. The symptoms respond to drainage of CSF either by a needle in the spine (a "lumbar puncture") or by a permanently implanted drain (a CSF "shunt"). The basis for this problem is not well understood, but the leading hypothesis is that both the CM1 and the pseudotumor are caused by an obstruction of cerebral venous drainage.

Even if it successfully relieves all symptoms, surgical treatment for the CM1 should not be considered a cure, especially if there is associated syringomyelia. The long-term outlook for treated patients is really not known. Periodic neurosurgical reassessment and, at the surgeon's discretion, periodic MR imaging are appropriate to monitor for complications.

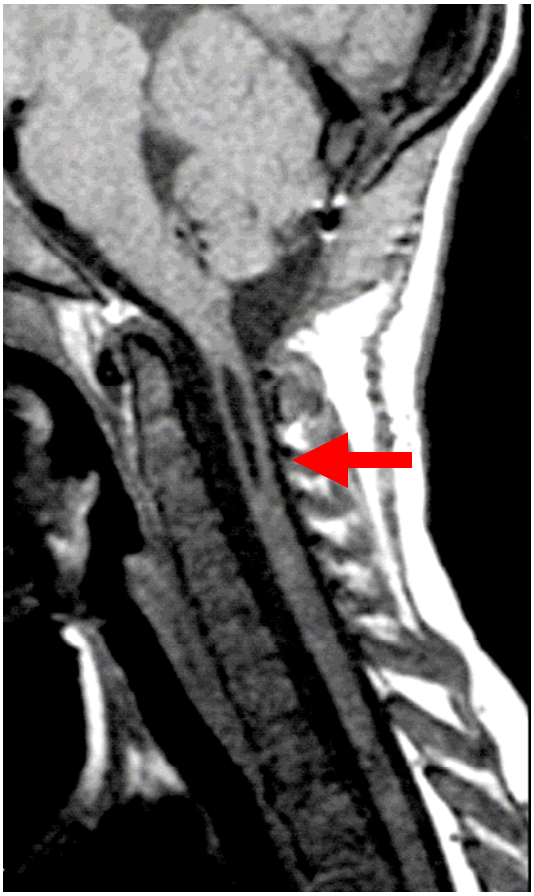

Figure 4. *This sagittal MR image represents the same patient depicted in Figure 2 after surgical treatment. In this sequence the CSF is black. The red arrow marks the syrinx cavity that was distended in Figure 2. Now it is collapsed; it no longer widens the silhouette of the spinal cord.*

Drexel University College of Medicine Pediatric Neurosurgery HOME

warning!