YOUR REFERENCE BOOKLET

A Comprehensive Guide to Understanding the Chiari I Malformation and Syringomyelia

Providing Research, Education and Support for Syringomyelia and Chiari Malformation

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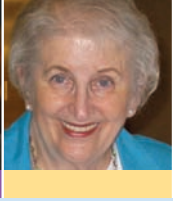
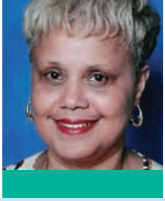
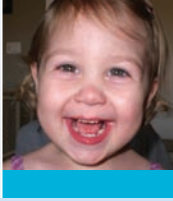

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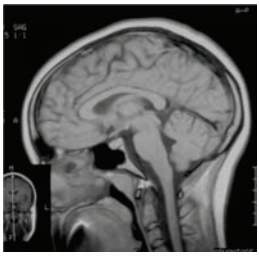
Chiari Malformation

What is the Chiari malformation?

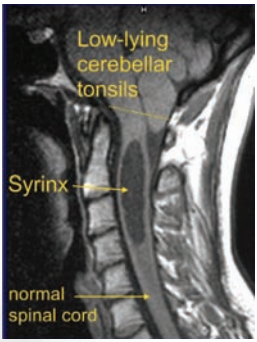
The Chiari malformation is an abnormality in the lower part of the brain called the cerebellum. There are several different forms. The most common type is the Chiari I malformation (CM) which this article addresses. Less commonly, it may be known as Arnold-Chiari malformation, tonsillar herniation or tonsillar ectopia. Most cases of Chiari are congenital, meaning they are present from birth.

To help explain this condition, a quick lesson in brain anatomy is helpful.

In normal anatomy, the cerebellar tonsils are located just above the line called the foramen magnum. But in an individual with Chiari, the tonsils hang below the line (herniate) into the spinal canal. The degree to which the tonsils extend can vary tremendously.



Normal brain and spinal cord



An individual with the Chiari malformation and a syrinx

What are the symptoms?

The most common symptom of Chiari malformation is a headache, which begins at the back of the head (neck) and radiates upward. The pain is often made worse or can be brought on by coughing, sneezing or straining. These activities are known as valsalva maneuvers. A valsalva maneuver compresses the blood flow in the chest, which in turn causes increased pressure in the head. This can bring on severe symptoms in an individual with Chiari.

Visual problems such as nystagmus (involuntary eye movements), double or blurred vision may occur. Balance difficulties, vertigo and dizziness also may be present. Some people may have cranial nerve compression. This can result in apnea (lapse in breathing), gagging, swallowing difficulties, facial numbness or syncope (temporary loss of consciousness).

Patients may have muscle weakness, particularly in the upper extremities, coordination problems, or gait abnormalities. Imaging of the spine may reveal a fluid collection inside of the spinal cord, known as a syrinx. Some individuals may have hydrocephalus, a buildup of fluid in the ventricles of the brain.

Very young children often show a specific group of symptoms: failure to thrive, repeated incidents of aspiration pneumonia or a diagnosis of gastrointestinal reflux, difficulty swallowing, sleep apnea, developmental delays and/or scoliosis.

It is possible to have a Chiari malformation and have no symptoms. In fact, Chiari malformations have been noted incidentally on MRIs taken for different reasons. Researchers are looking for clues to help them better understand why some people have symptoms and others do not.

Researchers have found that the degree of herniation does not always correspond with the severity of symptoms. Some mild herniations produce large syrinxes or severe symptoms, while large herniations may cause no symptoms at all.

What is the treatment?

The first step after diagnosis is to consult with a neurosurgeon who has experience treating and managing this disorder. Be aware that you may need to travel and you may wish to consult with more than one specialist.

If symptoms are mild and not progressing, your doctor may recommend conservative management. Supportive care such as headache and pain management, physical therapy or a reduction in activities can help manage symptoms.

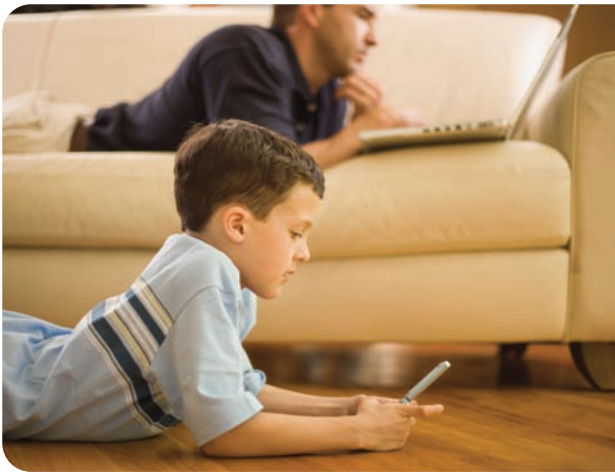
An operation may be recommended. This is referred to as a posterior fossa decompression. The surgeon makes more room in the back of the head by removing small pieces of the skull bones. This reduces compression of the brain stem and allows the tonsils to move back into their natural position. The specific surgical techniques will vary among surgeons; no consensus yet exists on the best variation on this surgical procedure.

An average hospital stay ranges from 3 to 5 days. The first night after surgery is usually spent in the intensive care unit. The typical patient is able to return to full activities within 4 to 6 weeks of surgery. Depending upon your individual situation, some restrictions on activities could be necessary. Physical therapy may be recommended to help regain function and strengthen neck muscles.

What is the prognosis?

The prognosis depends on the patient's overall health prior to surgery and the presence of other conditions. While not everyone's symptoms will completely go away, most patients will notice improvement. In some cases, additional surgical procedures could be necessary.

When surgery is not recommended, treatment plans are typically guided by regular neurological exams along with MRI scans. Symptoms could progress, stay the same, or improve over time.



Is Chiari hereditary?

Researchers investigated the genetic implications of the Chiari malformation with or without syringomyelia. A genetic prevalence has been identified in some families. Researchers continue to search for the gene(s) that are responsible for producing the Chiari malformation.

MRI scanning is recommended for family members who have signs or symptoms of the disorder.

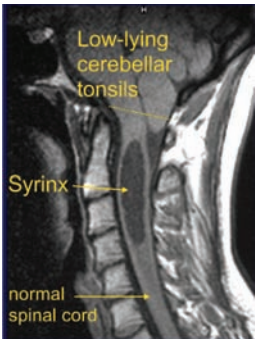
Syringomyelia

What is a syrinx?
What is syringomyelia?

The brain and spinal cord are surrounded by cerebrospinal fluid (CSF). This fluid helps transport nutrients, eliminates waste products, and provides a cushion for the brain and spinal cord from trauma.

When the normal flow of spinal fluid is disrupted or blocked, it can be forced inside the spinal cord. The way in which this occurs is not completely understood. As spinal fluid is forced into the spinal cord a fluid-filled cavity is created. This is known as a syrinx. It is also sometimes called hydromyelia. Syrinxes are most commonly found in the cervical spinal cord but can also be found in the thoracic and/or lumbar regions.

Over time, the syrinx can elongate or widen. This damages the spinal cord and can result in serious and/or disabling symptoms. The condition of having a syrinx is known as syringomyelia (SM).



Note the dark area of the spinal cord. That is the syrinx.

What causes syringomyelia?

The most common cause of SM is a congenital condition known as the Chiari malformation (CM). The cerebellar tonsils lie abnormally low in the spinal canal, causing a blockage in the normal flow of spinal fluid.

The second major cause of a syrinx is spinal cord injury. Bony abnormalities, hemorrhaging and scarring that result from severe spinal damage often contribute to a syrinx development. This may occur years after the original injury. The syrinx can cause further muscle weakness or additional loss of function which can be devastating to a spinal cord injury patient.

SM can be caused by any condition that blocks the normal flow of spinal fluid. Other causes include spinal or brain tumors, arachnoid cysts, meningitis (which can cause arachnoiditis or scarring), a tethered spinal cord, a condition in which the cord lies lower than normal, prohibiting it from moving freely.

What are the symptoms?

Symptoms develop slowly over time, but can come on suddenly after a fall or minor trauma. Sensation may be affected first. Loss of sensitivity to hot and cold, numbness and tingling can occur. Bowel and bladder function may be affected. Motor symptoms include muscle weakness, spasticity, paralysis and, in severe cases, quadriplegia.

Scoliosis is a common finding in individuals with SM, and it may be the only symptom in children. Often people with scoliosis undergo spinal MR imaging because of an atypical left-sided thoracic curve.

Many individuals suffer from chronic pain and some will develop neuropathic pain syndromes. This type of pain is difficult to treat. A large percentage of people have headaches which can be severe.

What is the treatment?

The first step after diagnosis is to consult with a neurosurgeon who has significant experience in treating and managing this disorder. Be aware that you may need to travel and you may wish to consult with more than one specialist.

Treatment is aimed at restoring the normal flow of spinal fluid. In persons with Chiari and SM, a procedure known as a posterior fossa decompression may be recommended. This procedure allows the cerebellar tonsils to move into a normal position, restoring the normal flow. After this procedure the syrinx will often reduce or resolve on its own. This can take months and may never completely collapse the syrinx. Some patients may require repeat surgeries. The most important consideration in deciding to undergo surgery is the severity of symptoms and overall quality of life issues.

In syrinxes associated with other causes, the surgical treatment is also aimed at restoring normal physiology and flow of spinal fluid. This might involve a bony spinal decompression, removing a tumor or scar tissue. In cases of tethered spinal cord, a surgical procedure is used to release the tension on the cord, allowing it to return to its normal position.

The most desirable outcome of any surgery for SM is to stop progression. Surgery may improve symptoms and reduce the size of the syrinx. However, damage to the spinal cord is not always reversible and neurological pain and dysfunction may not completely go away. SM associated with CM usually has a better surgical outcome than other causes of SM. Children with Chiari and syringomyelia, in particular, tend to have the best outcomes. That is why early diagnosis is essential.

Syrinx drainage used to be the primary treatment of SM. In a shunting procedure, a tiny tube is inserted into the syrinx which

drains to a cavity outside of the spinal column. A subarachnoid shunt drains the syrinx fluid into the space surrounding the spinal cord.

Shunting procedures are associated with a greater risk. Shunts frequently become closed or dislodged, and repeated surgery may be necessary. In addition this procedure does not address the underlying reason the syrinx developed.

Not all patients will require surgery. If symptoms are mild and not progressing, your doctor may recommend conservative management. Non-surgical interventions such as headache management, pain medication, physical therapy, occupational therapy and changes in lifestyle can help manage symptoms.

Is the condition progressive?

The natural history of untreated SM is poorly understood. Some patients will continue to have a progression of symptoms, and the syrinx cavity will continue to enlarge. Others will remain stable for years or a lifetime. Regular neurological exams and periodic MRI scans help monitor your condition.

Special considerations for children with CM and/or SM

Special considerations for children with syringomyelia and/or Chiari

When a child is diagnosed it affects the whole family, not just the child. Parents may feel overwhelmed, powerless, and afraid. But there is a lot you can do. Taking control and playing an active role in your child's healthcare is essential. By researching the condition, you will be better prepared to advocate for your child.

Children and adolescents should be treated by a pediatric neurosurgeon. Generally pediatric neurosurgeons are affiliated with children's hospitals. Regular monitoring is critical when dealing with children who may not be able to communicate symptoms as well as adults can.

A pediatric neurosurgeon will perform a detailed exam and review of films. Additional testing may be necessary. Other specialists may need to be consulted, i.e., orthopedist, neuro-ophthalmologist, urologist, neurologist.

Children who have a functional impairment will qualify for accommodations to help them succeed in school. An individual educational plan (IEP) can be developed with the help of your school.

Activities

Whether or not your child has had surgery, there are no general rules pertaining to permitted activities. However, this is an important topic to discuss with the pediatric neurosurgeon who cares for your child. Some children who have had extensive surgical procedures or who have bony abnormalities may be prohibited from certain activities.

We know that symptoms can be brought on by a fall or minor trauma. Be sure your child wears protective head gear when participating in activities such as riding a bike or a scooter.

The asymptomatic child

Due to the widespread availability of MRI technology, more children are being scanned now than ever before, and in some cases, a Chiari malformation is discovered despite having no symptoms.

Learning that your child has Chiari and/or a syrinx when you had no idea that anything could be wrong is a frustrating and worrisome situation for parents. On the one hand, parents do not want their child to undergo surgery, but they fear without surgery the child could suffer from permanent damage in the future. The natural history of Chiari with or without syringomyelia is not fully understood. Some people may go their entire lives and never develop any symptoms. Therefore, surgery is rarely recommended for the asymptomatic child. Nevertheless, a pediatric neurosurgeon should be consulted in all cases.

Talk to your child

Children may feel isolated and different from their peers when they are dealing with a disorder such as CM or SM. Letting children talk about their feelings will help them cope. They may have unfounded fears and worries such as dying or becoming paralyzed.

It is important to explain the condition and necessary procedures in a way your child can understand. Consider discussing the details of surgery with the surgeon while the child is not in the room. However, encourage the child to voice questions to the neurosurgeon. Most children's hospitals have child life specialists who can help with the process of preparing for surgery.

Related disorders

Related disorders

There are several conditions that are often associated with Chiari and syringomyelia. They include:

Arachnoiditis—inflammation of the arachnoid membrane in the spinal canal. It can be caused by trauma, spinal surgery, hemorrhage, or infection. It can result in severe pain. There is no cure or treatment when it is widespread.

Basilar invagination—a condition in which a portion of the C2 to vertebra (called the odontoid), compresses the brain stem. It is diagnosed with x-rays and MRIs.

Chiari II malformation—usually found in children with spina bifida or myelomeningocele. Not only is part of cerebellum unusually low and lying below the bottom of the skull, but the brain stem can be malformed in several ways.

Chiari III and IV malformation—Types III and IV represent gross herniations of the cerebellum and are very rare. They represent total herniation of the brainstem and cerebellum into the spinal canal, and are often associated with a cervical encephalocele.

Hydrocephalus—a buildup of fluid in the ventricles of the brain. This condition is usually treated by a placing a ventricle-peritoneal shunt to drain the fluid into the peritoneal (lining surrounding contents of the belly) cavity. The condition can be diagnosed by CT scan.

Intracranial hypertension (pseudotumor cerebri)—an abnormally high cerebrospinal fluid (CSF) pressure in the brain. Symptoms include headache, nausea, vomiting and visual problems. Optic nerve swelling can be diagnosed by a dilated eye exam. Pressure is measured by spinal tap, but more extensive monitoring may be necessary in a hospital setting. In these cases, a neurosurgeon will drill a small hole in the skull and insert a fiber optic wire into the skull to measure the CSF pressure for an extended period of time. This gives the surgeon a more accurate clinical picture. The wire is then removed, and generally only one staple or a single stitch is needed to close the wound. Treatments include medical therapies to reduce CSF fluid production or a shunt to remove CSF.

Neuropathic pain syndrome—pain caused by damage to the central nervous system. This is characterized by burning pain and abnormal sensations. It can be associated with allodynia, which is a painful sensation occurring after light touch. Neuropathic pain is difficult to treat but responds best to the drugs that specifically treat neuropathic pain.

Spina bifida—a birth defect in which the spinal cord fails to close during fetal development. There are many different forms of spina bifida ranging from benign (spina bifida occulta) to severe (myelomenigocele). Most patients with myelomeningocele will have the Chiari II malformation.

Syringobulbia—the formation of a syrinx in the brain stem.

Chiari malformation (CM) and syringomyelia (SM) at a glance

- Estimates are that syringomyelia affects over 250,000 Americans. No one knows how many people have Chiari, but the number is likely to be at least double that of SM.
- CM and SM are diagnosed by MRI.
- Most cases of Chiari are detected in early adulthood, but as awareness and technology increase, children are diagnosed more frequently.
- Chiari is thought to be congenital but can be acquired due to conditions such as hydrocephalus, intracranial hypertension and certain shunt types such as the lumboperitoneal shunt.
- Chiari is the most common cause of syringomyelia, but not every person with Chiari will develop a syrinx.
- Idiopathic syrinxes—those with no known cause—are uncommon. A thorough search for the cause should be undertaken.
- Chiari I malformation and syringomyelia are not considered fatal conditions.
- Not all patients need surgery. The decision to operate requires careful consultation between the patient and surgeon and is usually based on symptoms.
- A syrinx can resolve on its own without any medical therapy, although this is very rare.
- Tonsillar herniation can increase over time or after trauma. Alternately, the degree of herniation can recede. This is more common in young children whose growing skulls may permit more room for the tonsils.
- Many individuals suffer from chronic pain and headaches, but not all.
- Surgical treatment for SM is aimed at restoring the normal flow of spinal fluid.
- Although surgery may not result in the complete disappearance of a syrinx, a successful surgery will, in most cases, allow the syrinx to reduce and stop progressing.

About ASAP

About ASAP

The American Syringomyelia Alliance Project, Inc. (ASAP) is a not-for-profit, 501(c)(3), member-supported organization chartered in May 1988. ASAP is the result of the commitment and determination of its two founding members, Barbara and Don White, whose journey with syringomyelia (SM) and Chiari malformation (CM) began in1983.

Barbara was experiencing numbness in her left hand, headaches and other unexplained symptoms. After three weeks of hospitalization and testing, she was finally diagnosed with syringomyelia. Barbara later learned she also had Chiari.

Their efforts to learn about these conditions proved to them that very little was known, and few physicians were even aware of the limited information that was available. The frustrations they faced years ago mirror those of many with SM and CM even today. While we now know that more than 70% of SM cases are caused by Chiari malformation, much is still unknown. ASAP's goal is to provide people with SM/CM the support and direction that was unavailable to Barbara when she began her journey. Unfortunately, she did not live to see the blossoming of her organization. Barbara died from complications of syringomyelia and Chiari in 1991. However, ASAP continues to make strides toward the goals she set.

ASAP's Mission
Help Us

To improve the
lives of persons affected
by syringomyelia,
Chiari malformation and
related disorders while
we find the cure

ASAP provides:

- *Connections*, a bi-monthly newsletter
- An annual conference
- Funding for research
- Teleconferences and webinars
- Networking and peer support
- An informational web site
- Email announcements
- Educational materials
- Awareness merchandise

This brochure has been prepared by
the American Syringomyelia
Alliance Project, Inc.

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