

Chiari I Malformation & Syringomyelia

Overview

Chiari (pronounced key-AR-ee) malformation is a condition in which the lower part of the brain, called the cerebellar tonsil, herniates down through the skull and into the spinal canal. The herniated tissue blocks the normal flow of cerebrospinal fluid (CSF). Instead of moving in an easy, pulsating movement through this opening, the fluid begins to force its way through – like a water hammer – pushing the tonsils down even farther. The blockage can cause a buildup of fluid in the spinal cord (syringomyelia) or in the brain (hydrocephalus).

Chiari is often misdiagnosed because the wide variety of bony and soft tissue problems can cause a wide array of possible symptoms (headache, neck pain, dizziness, arm numbness or weakness, sleep problems, etc.) Headache in the back of the head that worsens with coughing, sneezing, or straining is a hallmark sign.

Treatment options depend on the type of Chiari and the severity of symptoms. If symptoms are mild, regular monitoring and medications can be effective. However, symptoms typically progress and worsen over time. Surgery may be performed to remove a part of the skull bone and create space for the cerebellum and brainstem. An accurate diagnosis and prompt treatment are important to prevent permanent injury to the nervous system.

Types of Chiari

Chiari type I, the most common, affects both children and adults. Because the back of the skull is too small or deformed, a crowding of the brainstem, cerebellum, and tonsils occurs (Fig. 1). As the tonsils push out of the skull, they block CSF flow. Chiari I may cause a fluid-filled cyst (syrinx) in the spinal cord.

Chiari type 0, a newly identified form of Chiari, describes the absence (or a “zero” herniation) of the tonsils below the foramen magnum. Yet Chiari 0 includes the presence of both symptoms and a syrinx in the spinal cord. This new type is under study and controversial.

Chiari type II is present at birth and affects infants. It occurs with the birth defect myelomeningocele, a form of spina bifida. When the spinal canal does not close before birth, some of the spinal cord protrudes like a sac from the baby’s

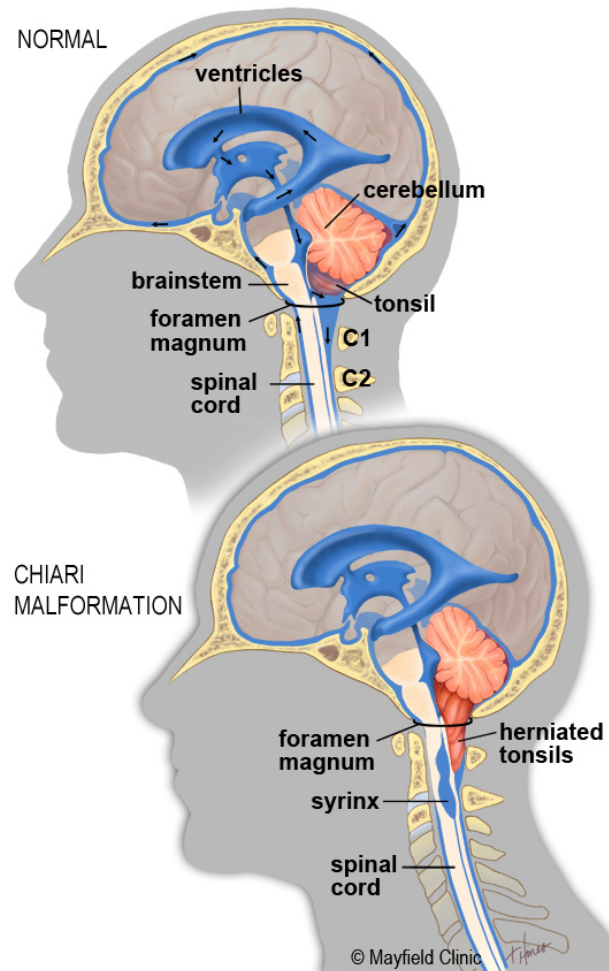


Figure 1. Normal anatomy of the cerebellum compared to Chiari I malformation. The posterior fossa is too small causing the cerebellar tonsils to herniate through the skull (foramen magnum) into the spinal canal. The tonsils block the flow of CSF (blue) and may cause fluid buildup inside the spinal cord, called a syrinx.

back. Both the brainstem and tonsils are pulled down into the spinal canal to block CSF flow in the brain and causing hydrocephalus. This type is correctly called Arnold-Chiari malformation.

Chiari type III affects infants and is a rare but severe herniation that involves the cerebellum. It can develop with the birth defect encephalocele, a fluid-filled sac at the back of the baby’s neck.

Chiari type IV affects infants. This rare and often fatal malformation occurs when the cerebellum does not develop properly.

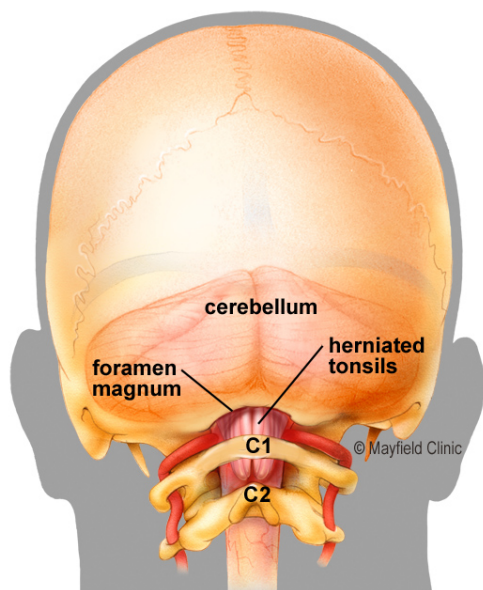


Figure 2. The tonsils are pushed out of the skull through the foramen magnum into the spinal canal.

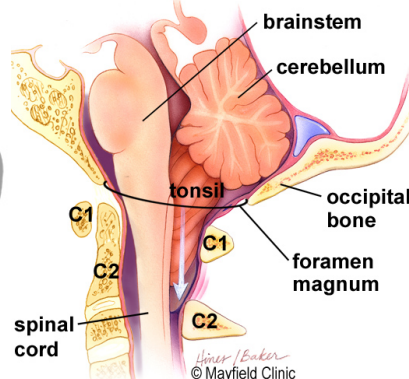


Figure 3. With each heartbeat, the tonsils may "piston" up and down blocking CSF flow.

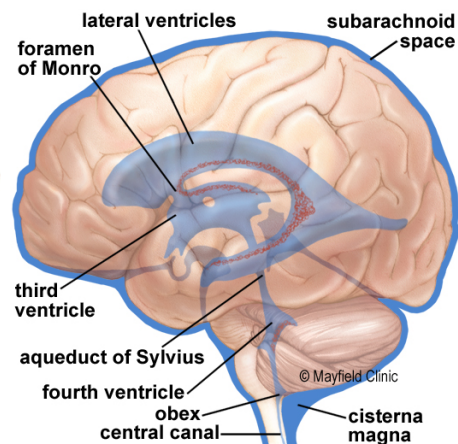


Figure 4. Cerebrospinal fluid (CSF) circulates through the ventricles inside the brain to the spaces surrounding the brain and spinal cord.

What is a Chiari I malformation?

Chiari I begins with the underdevelopment of the fetal skull during pregnancy. During childhood, the brain continues to grow and the skull hardens. However, the small size or shape of the Chiari skull is mismatched to the size of the brain. Thus, a crowding of the brainstem, cerebellum, and tonsils occurs. Crowding pushes the tonsils out of the skull through the opening (foramen magnum) where the spinal cord exits (Fig. 2). Herniation of the cerebellar tonsils can extend several millimeters below the foramen magnum. The tonsils put pressure on the brainstem and spinal cord, block CSF flow, and result in Chiari signs and symptoms.

Bone deformity

A variety of bone abnormalities can occur in Chiari:

- The posterior fossa may be smaller than normal. If too small, the effects can be crowding of the brainstem and cerebellum, as well as herniation of the tonsils through the foramen magnum.
- The occipital bone may be misshaped or thickened.
- Basilar invagination occurs when the top of C2 (odontoid) pushes upward into the brain. This defect can narrow the foramen magnum and crowd the brainstem and cerebellum.
- Scoliosis is a curvature of the bony spine. There is a high rate of scoliosis with Chiari and syringomyelia, especially in children.
- Ehlers-Danlos syndrome (EDS) is a connective tissue disorder that may increase the incidence and severity of Chiari. EDS causes joint hypermobility and loose/unstable joints. At the craniocervical junction, strong ligaments attach the C1 and C2 vertebrae to the skull, allowing

movement of the head. For someone with both Chiari and EDS, extra testing and precautions are taken to ensure the connection between the spine and skull is intact. Spinal fusion surgery may be needed to support the neck and skull.

Cerebellum herniation

The cerebellum is the lower part of the brain located in the posterior fossa. On the underside of the cerebellum are two tonsils. The cerebellum coordinates body movement. It maintains muscle tone and balance. The cerebellum is also involved in attention, language, memory, and learning. Signs of cerebellum problems include loss of coordination, unstable walking (gait), trouble with speech, and difficulty with eye movement and swallowing.

In Chiari, the cerebellar tonsils are stretched as they push through the foramen magnum into the spinal canal (Fig. 3). This results in compression of the brainstem and spinal cord. The extent of tonsillar herniation can be seen on MRI.

Brainstem compression

Acting as a relay center, the brainstem connects the cerebrum and cerebellum to the spinal cord. The brainstem performs many automatic functions such as breathing, heart rate, body temperature, wake and sleep cycles, digestion, sneezing, and coughing. Ten of the 12 cranial nerves originate in the brainstem.

In Chiari, compression of the brainstem and cranial nerve nuclei can occur. Patients may experience problems with sleeping (pons), breathing (medulla), swallowing, facial pain or numbness, hearing loss, irregular heartbeat, and digestion.

Cerebrospinal fluid blockage

Cerebrospinal fluid (CSF) is a clear, watery-like liquid that flows within and around the brain and spinal cord to help cushion it from injury. This fluid is produced inside the ventricles by the choroid plexus and is constantly being absorbed and replenished.

The CSF flows through the ventricles and out into the space between the brain and skull (subarachnoid space) and down into the spinal canal (Fig. 4). A large amount of CSF lies at the back of the cerebellum in an area called the cisterna magna.

As the heart beats, CSF flows into the brain. This is normally balanced by CSF then flowing from the brain into the spinal compartment. In a Chiari malformation, this balanced flow is disrupted. The obstructed CSF begins to force its way like a water hammer through the foramen magnum. Pushing the tonsils down even farther, it exerts pressure on the brainstem. The increasing pressure compromises normal functions of the brain and/or spinal cord and a myriad of symptoms occur.

Excess CSF can collect and enlarge either the ventricles in the brain (hydrocephalus), or form a cyst in the spinal cord (syringomyelia).

Syringomyelia, hydrocephalus, and other complications. When cerebrospinal fluid (CSF) flow is obstructed and collects within the spinal canal, it can eventually form a syrinx. This condition, called syringomyelia (pronounced sir-RING-o-my-el-ia), damages the spinal cord. The compressed nerve fibers inside the cord cause a wide variety of symptoms. Problems affect the arms or legs, or affect feeling, strength, or balance. Syringomyelia occurs in about 65% of patients with Chiari I malformation.

In some cases, the CSF collects within the ventricles of the brain (hydrocephalus); this condition may require placement of a shunt to divert this excess fluid. Bony abnormalities, which affect about 25% of patients, can include basilar invagination, scoliosis, and cranial cervical instability.

What are the symptoms?

Chiari I symptoms vary from person to person and are not necessarily related to the size of tonsillar herniation. Some people with large herniations have no symptoms (asymptomatic). Yet others with small herniations have severe symptoms. When symptoms are present, they are often vague or nonspecific. As a result, the diagnosis of Chiari is often delayed until more severe symptoms occur or after current symptoms persist for some time. Symptoms are caused by disruption of the CSF flow and compression of nervous tissues.

Chiari I symptoms

Pressure-like headaches at back of skull
Headaches worsen with coughing, sneezing
Neck and shoulder pain
Ringing or buzzing in the ear (tinnitus)
Dizziness, vertigo
Trouble walking (gait), imbalance
Difficulty swallowing, gagging
Facial pain, numbness, or tingling
Hoarseness, change in voice
Snoring / sleep apnea
Fatigue / insomnia
Problems with memory / concentration
Nervousness / anxiety / depression
Trouble speaking, word finding
Blurred or double vision
Jerking eye movements (nystagmus)
Difficulty tracking or following objects
Irregular heart beat
Black out spells / syncope

Syringomyelia symptoms

Headaches (due to Chiari malformation)
Loss of sensitivity, especially to hot and cold
Muscle weakness and spasticity
Numbness in hands and feet
Pain in neck, arms and back
Loss of bowel and bladder control
Scoliosis

Because the brainstem is responsible for most body functions, Chiari causes all kinds of strange symptoms. People may experience symptoms that range from headache to irritable bowel. The five most common symptoms are:

1. Pressure-like headaches at the back of the skull that worsen with physical strain or coughing; often with neck pain
2. Hoarseness or swallowing problems
3. Sleep apnea
4. Weakness or numbness in an extremity
5. Balance problems

People with Chiari I often develop symptoms during their teen or early adult years. The disorder is also seen in young children and older adults. In some cases, a head or neck injury from a car accident or sports injury triggers the onset of symptoms.

Who is affected?

Chiari I is seen on MRI scans in people of all ages. Its incidence was earlier estimated to affect 1 in every 1,000 births. Now with increasing use of diagnostic imaging, Chiari may be far more common. Patients typically seek medical attention in their 20s and 30s. Three times more women than men are affected. Genetic studies show that Chiari may cluster in some families.

How is a diagnosis made?

The complex symptoms of Chiari I can mimic other diseases – often leading to misdiagnosis and delay in treatment. At times, Chiari I is mistaken for fibromyalgia, chronic fatigue syndrome, migraine, multiple sclerosis, mental disorder, depression, sinus disease, trigeminal neuralgia, or other neurologic disorders.

There is no specific test to confirm Chiari. Rather, a diagnosis is made by assessment of the patient's symptoms, neurological exam, and MRI findings.

A complete medical history and physical exam can determine if your symptoms are related to Chiari or another problem. A neurological exam detects problems with cranial nerves such as gag reflex, facial numbness, hoarseness, double vision, tremors, and vision problems. You may be asked to see an eye or ear specialist, or to undergo a sleep evaluation. Your doctor will order one or more imaging studies, including:

Magnetic resonance imaging (MRI) scan is a noninvasive test used to evaluate the brain, spinal cord, and surrounding CSF. MRI can identify the extent of cerebellar herniation (Fig. 5). The herniation may reach to the level of the first two vertebra (C1 or C2) of the cervical spine. Herniation of the tonsils is often measured in millimeters (mm) below the foramen magnum. The classic definition of Chiari I is herniation greater than 5mm below the foramen magnum. However, the size of herniation seen on MRI does not closely correlate with symptoms. Someone without herniation may have severe symptoms while another with 20-mm herniation may have no symptoms. MRI of the spine can detect abnormal accumulations of CSF within the spinal cord (Fig. 6). This fluid-filled cavity (syrinx) is surrounded by stretched tissues of the spinal cord.

Cine MRI scan is a special study used to observe cerebrospinal fluid (CSF) flow. With each heartbeat, CSF is forced out of the ventricles of the brain, into the cisterna magna, and down the spinal canal. When the heart relaxes, the CSF flow reverses. The movie-like cine MRI captures the fluid movement. The test can determine if, and by how much, a Chiari is blocking the back-and-forth flow of CSF between the brain and spine.

Computed tomography (CT) scan is used to view the bony skull base and spinal column. It can detect thickened bone or previous trauma.

X-rays of the neck may be taken in flexion and extension to view the bony vertebrae. These images can help your doctor identify any instability at the craniocervical area.

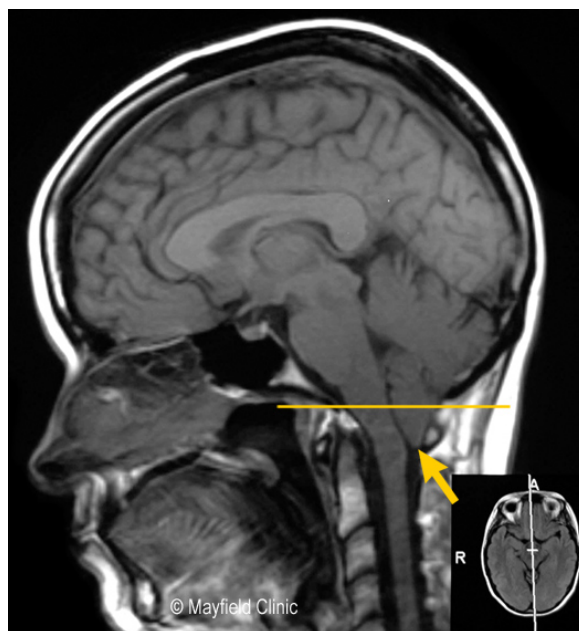


Figure 5. An MRI of the brain shows the cerebellar tonsils (arrow) herniating through the foramen magnum (yellow line).



Figure 6. An MRI of the neck shows a collection of CSF in the spinal cord (yellow arrow) called a syrinx.

What treatments are available?

Treatment options vary depending on the severity of symptoms, the extent of tonsil herniation, and the presence of other conditions such as syringomyelia.

Observation (watch and wait)

Monitoring by regular check-ups and periodic MRI scans may be recommended for those with mild or no symptoms. Headache can be relieved with anti-inflammatory or pain-relieving drugs.

Follow these self-care tips to minimize neck strain in daily activities:

- Ice packs for 20 minutes can help relieve neck and shoulder pain.
- Get at least 8 hours of sleep and use a good pillow.
- Have a sleep study and evaluation for sleep apnea. A CPAP (Continuous Positive Airway Pressure) machine can greatly improve your sleep quality and reduce fatigue.
- If you are overweight, shed extra pounds to reduce the strain on your arms and legs and help with numbness/tingling sensations.
- Stay active with low-impact activities, such as walking, cycling, or water aerobics.
- Play cards, crossword or Sudoku puzzles to sharpen your thinking.
- Tai Chi or yoga can help stretch and tone muscles, improve balance, and reduce stress. Avoid poses that aggravate your symptoms.

Avoid these activities if you have a Chiari:

- High-velocity chiropractic manipulation can worsen the herniation and injure the spinal cord.
- Cervical traction.
- Trampolines, roller coasters, scuba diving, and other activities that apply G forces to the neck.
- Contact sports to avoid: football, soccer (heading the ball), diving, running, weight lifting, etc.
- Constipation and straining during bowel movements. Straining can cause formation or worsening of a syrinx.
- Lumbar punctures (spinal taps) and epidurals can be dangerous and increase the herniation.

Childbirth (bearing down and pushing) can also increase cerebellar herniation and formation of a syrinx. Make sure your OB/GYN is aware of your Chiari and tell your neurosurgeon if you become pregnant.

It is important for patients to closely monitor their symptoms. Some patients find it helpful to keep a **symptom diary**. By keeping track daily of how you feel and what you do, you may be able to find patterns, identify triggers, and notice subtle changes over time. Bring the symptom diary to

each appointment to help you communicate more clearly with your doctor. Knowing what symptoms you experience most, and to what degree, can help shape your diagnosis and treatment. If your symptoms worsen or if any new ones develop, tell your neurosurgeon promptly.

Surgery

Surgery is advised for those with moderate to severe symptoms or with a syrinx. The goals of surgery are to stop or control the progression of symptoms caused by herniation of the cerebellar tonsils, and relieve compression of the brainstem.

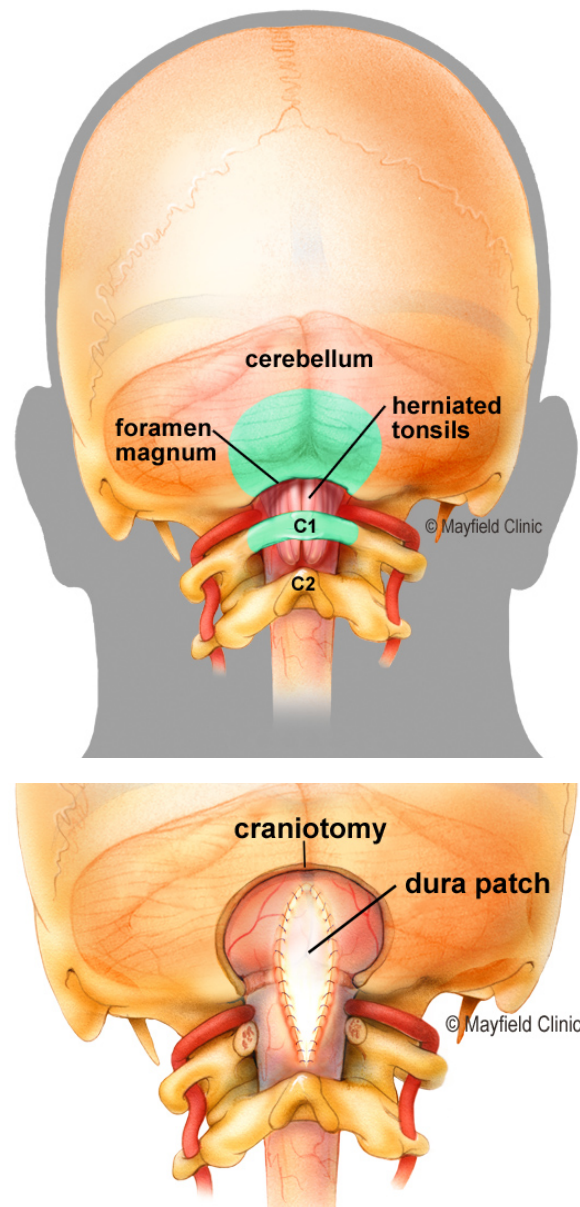


Figure 7. Posterior fossa decompression surgery removes bone (green areas) and creates more space for the brainstem and cerebellum. The dura is opened and a patch is sewn to enlarge the CSF space.

Posterior fossa decompression surgery removes bone (craniectomy) at the back of the skull and spine to widen the foramen magnum. The surgeon opens the dura overlying the tonsils and sews a dura patch to expand the space, similar to letting out the waistband on a pair of pants (Fig. 7). After surgery, symptoms related to the blockage of CSF should decrease as flow normalizes.

Spinal fusion may be performed in addition to decompression surgery in certain patients with spine instability. The neck area of the spine may be unstable due to scoliosis, Ehler-Danlos syndrome, or other bone abnormality. Rods and screws are inserted to reinforce the skull and neck vertebrae.

Shunting is used to reroute CSF. The shunt includes a flexible tube with a 1-way valve that directs the fluid out in the desired direction. For a syrx in the spinal cord, one end of the tubing is placed in the syrx. The other end is placed outside the spinal cord. For hydrocephalus, one end of the tubing is placed in the ventricle of the brain. The other end is placed in the abdomen (called a ventriculoperitoneal shunt). A shunt remains inside the body after surgery. However, shunts pose risks and often become clogged or dislodge. Repeated surgeries may be necessary.

Transoral decompression is a surgical procedure to treat basilar invagination. The surgery is performed through the mouth and to the back of the throat to remove an abnormal odontoid bone (C2 vertebra).

Clinical trials

Clinical trials are research studies in which new treatments—drugs, diagnostics, procedures, and other therapies—are tested in people to see if they are safe and effective. Research is always being conducted to improve the standard of medical care. Information about current clinical trials, including eligibility, protocol, and locations, are found on the Web. Studies can be sponsored by the National Institutes of Health (see www.clinicaltrials.gov) as well as private industry and pharmaceutical companies (see www.centerwatch.com).

Sources & links

If you have more questions, please contact New England Neurological Associates at 978-687-2321.

Links

www.asap.org
www.conquerchiari.org
www.ehlers-danlos.com

Glossary

basilar invagination: a condition in which the upper portion of the second cervical vertebra (C2) migrates upward and back into the intracranial space.

cerebrospinal fluid (CSF): a clear fluid produced by the choroid plexus in the ventricles of the brain. CSF bathes the brain and spinal cord, giving them support and buoyancy to protect from injury.

craniectomy: surgical removal of a portion of the skull.

Ehler-Danlos syndrome (EDS): a rare genetic defect in collagen that affects connective tissue (e.g., joints, skin, blood vessels). Collagen is a protein, which acts as a "glue" in the body, adding strength and elasticity to connective tissue. There are six types of EDS. Types I - III cause joint hypermobility; joint dislocations and scoliosis are common.

fibromyalgia (FM): a chronic pain illness characterized by widespread musculoskeletal aches, pain, and stiffness; soft tissue tenderness; fatigue; and disturbs sleep, memory, and mood. Many people with fibromyalgia also have tension headaches, temporomandibular joint (TMJ) disorders, irritable bowel syndrome, anxiety, and depression.

hydrocephalus: an abnormal build-up of cerebrospinal fluid usually caused by a blockage of the ventricular system of the brain. Increased intracranial pressure can compress and damage brain tissue.

multiple sclerosis: a chronic disease in which the body's immune system eats away at the protective sheath (myelin) surrounding nerves in the brain and spinal cord. Symptoms often come and go and vary widely depending on the affected nerve fibers.

scoliosis: an abnormal side-to-side curvature of the spine.

shunt: a drainage tube to move cerebrospinal fluid from inside the ventricles of the brain into another body cavity (e.g., abdomen).

syngomyelia: a chronic progressive disease of the spinal cord caused by an obstruction of normal cerebrospinal fluid (CSF) flow that redirects the fluid into the spinal cord to form a syrx.

syrinx: a cavity filled with cerebrospinal fluid (CSF) that expands and elongates over time, destroying the center of the spinal cord.

tethered cord syndrome: a condition of the spinal cord caused by an abnormal attachment or "tether" of the cord to the bones of the spinal canal. The spinal cord gets stretched and can become damaged. The filum terminale is a fibrous thread which connects the very bottom of the spinal cord to the coccyx bone.



updated > 11.2018

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