



Chiari Malformation

U.S. DEPARTMENT OF HEALTH
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National Institutes of Health

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What are Chiari malformations?

Chiari malformations are structural defects in the base of the skull and cerebellum, the part of the brain that controls balance. Normally the cerebellum and parts of the brain stem sit above an opening in the skull that allows the spinal cord to pass through it (called the foramen magnum). When part of the cerebellum extends below the foramen magnum and into the upper spinal canal, it is called a Chiari malformation (CM).

Chiari malformations may develop when part of the skull is smaller than normal or misshapen, which forces the cerebellum to be pushed down into the foramen magnum and spinal canal. This causes pressure on the cerebellum and brain stem that may affect functions controlled by these areas and block the flow of cerebrospinal fluid (CSF)—the clear liquid that surrounds and cushions the brain and spinal cord. The CSF also circulates nutrients and chemicals filtered from the blood and removes waste products from the brain.

What causes these malformations?

CM has several different causes. Most often it is caused by structural defects in the brain and spinal cord that occur during fetal development. This can be the result of genetic mutations or a maternal diet that lacked certain vitamins or nutrients. This is called primary or congenital Chiari malformation. It can also be caused later in life if spinal fluid is drained excessively from the lumbar or thoracic areas of the spine either due to traumatic injury, disease, or infection. This is called acquired or secondary Chiari malformation. Primary Chiari malformation is much more common than secondary Chiari malformation.

What are the symptoms of a Chiari malformation?

Headache is the hallmark sign of Chiari malformation, especially after sudden coughing, sneezing, or straining. Other symptoms may vary among individuals and may include:

- neck pain
- hearing or balance problems
- muscle weakness or numbness
- dizziness
- difficulty swallowing or speaking
- vomiting
- ringing or buzzing in the ears (tinnitus)
- curvature of the spine (scoliosis)

- insomnia
- depression
- problems with hand coordination and fine motor skills.

Some individuals with CM may not show any symptoms. Symptoms may change for some individuals, depending on the compression of the tissue and nerves and on the buildup of CSF pressure.

Infants with a Chiari malformation may have difficulty swallowing, irritability when being fed, excessive drooling, a weak cry, gagging or vomiting, arm weakness, a stiff neck, breathing problems, developmental delays, and an inability to gain weight.

How are CMs classified?

Chiari malformations are classified by the severity of the disorder and the parts of the brain that protrude into the spinal canal.

Chiari malformation Type I

Type 1 happens when the lower part of the cerebellum (called the cerebellar tonsils) extends into the foramen magnum. Normally, only the spinal cord passes through this opening. Type 1—which may not cause symptoms—is the most common form of CM. It is usually first noticed in adolescence or adulthood, often by accident during an examination for another condition. Adolescents and adults who have CM but no symptoms initially may develop signs of the disorder later in life.

Chiari malformation Type II

Individuals with Type II have symptoms that are generally more severe than in Type 1 and usually appear during childhood. This disorder can cause life-threatening complications during infancy or early childhood, and treating it requires surgery.

In Type II, also called classic CM, both the cerebellum and brain stem tissue protrude into the foramen magnum. Also the nerve tissue that connects the two halves of the cerebellum may be missing or only partially formed. Type II is usually accompanied by a myelomeningocele—a form of spina bifida that occurs when the spinal canal and backbone do not close before birth. (Spina bifida is a disorder characterized by the incomplete development of the brain, spinal cord, and/or their protective covering.) A myelomeningocele usually results in partial or complete paralysis of the area below the spinal opening. The term Arnold-Chiari malformation (named after two pioneering researchers) is specific to Type II malformations.

Chiari malformation Type III

Type III is very rare and the most serious form of Chiari malformation. In Type III, some of the cerebellum and the brain stem stick out, or herniate, through an abnormal opening in the back of the skull. This can also include the membranes surrounding the brain or spinal cord.

The symptoms of Type III appear in infancy and can cause debilitating and life-threatening complications. Babies with Type III can have many of the same symptoms as those with Type II but can also have additional severe neurological defects such as mental and physical delays, and seizures.

Chiari malformation Type IV

Type IV involves an incomplete or underdeveloped cerebellum (a condition known as cerebellar hypoplasia). In this rare form of CM, the cerebellum is located in its normal position but parts of it are missing, and portions of the skull and spinal cord may be visible.

What other conditions are associated with Chiari malformations?

Individuals with CM often have these related conditions:

- **Hydrocephalus** is an excessive buildup of CSF in the brain. A CM can block the normal flow of this fluid and cause pressure within the head that can result in mental defects and/or an enlarged or misshapen skull. Severe hydrocephalus, if left untreated, can be fatal. The disorder can occur with any type of Chiari malformation, but is most commonly associated with Type II.
- **Spina bifida** is the incomplete closing of the backbone and membranes around the spinal cord. In babies with spina bifida, the bones around the spinal cord do not form properly,

causing defects in the lower spine. While most children with this birth defect have such a mild form that they have no neurological problems, individuals with Type II Chiari malformation usually have myelomeningocele, and a baby's spinal cord remains open in one area of the back and lower spine. The membranes and spinal cord protrude through the opening in the spine, creating a sac on the baby's back. This can cause a number of neurological impairments such as muscle weakness, paralysis, and scoliosis.

- **Syringomyelia** is a disorder in which a CSF-filled tubular cyst, or syrinx, forms within the spinal cord's central canal. The growing syrinx destroys the center of the spinal cord, resulting in pain, weakness, and stiffness in the back, shoulders, arms, or legs. Other symptoms may include a loss of the ability to feel extremes of hot or cold, especially in the hands. Some individuals also have severe arm and neck pain.
- **Tethered cord syndrome** occurs when a child's spinal cord abnormally attaches to the tissues around the bottom of the spine. This means the spinal cord cannot move freely within the spinal canal. As a child grows, the disorder worsens, and can result in permanent damage to the nerves that control the muscles in the lower body and legs. Children who have a myelomeningocele have an increased risk of developing a tethered cord later in life.

- **Spinal curvature** is common among individuals with syringomyelia or CM Type I. The spine either may bend to the left or right (scoliosis) or may bend forward (kyphosis).

How common are Chiari malformations?

In the past, it was estimated that the condition occurs in about one in every 1,000 births. However, the increased use of diagnostic imaging has shown that Chiari malformation may be much more common. Complicating this estimation is the fact that some children who are born with this condition may never develop symptoms or show symptoms only in adolescence or adulthood. Chiari malformations occur more often in women than in men and Type II malformations are more prevalent in certain groups, including people of Celtic descent.

How are Chiari malformations diagnosed?

Currently, no test is available to determine if a baby will be born with a Chiari malformation. Since Chiari malformations are associated with certain birth defects like spina bifida, children born with those defects are often tested for malformations. However, some malformations can be seen on ultrasound images before birth.

Many people with Chiari malformations have no symptoms and their malformations are discovered only during the course of diagnosis or treatment for another disorder. The doctor will perform a physical exam and check the person's memory, cognition, balance (functions controlled by the cerebellum), touch, reflexes, sensation, and motor skills (functions controlled by the spinal cord). The physician may also order one of the following diagnostic tests:

- **Magnetic resonance imaging (MRI)** is the imaging procedure most often used to diagnose a Chiari malformation. It uses radio waves and a powerful magnetic field to painlessly produce either a detailed three-dimensional picture or a two-dimensional "slice" of body structures, including tissues, organs, bones, and nerves.
- **X-rays** use electromagnetic energy to produce images of bones and certain tissues on film. An X-ray of the head and neck cannot reveal a CM but can identify bone abnormalities that are often associated with the disorder.
- **Computed tomography (CT)** uses X-rays and a computer to produce two-dimensional pictures of bone and blood vessels. CT can identify hydrocephalus and bone abnormalities associated with Chiari malformation.

How are Chiari malformations treated?

Some CMs do not show symptoms and do not interfere with a person's activities of daily living. In these cases, doctors may only recommend regular monitoring with MRI. When individuals experience pain or headaches, doctors may prescribe medications to help ease symptoms.

Surgery

In many cases, surgery is the only treatment available to ease symptoms or halt the progression of damage to the central nervous system. Surgery can improve or stabilize symptoms in most individuals. More than one surgery may be needed to treat the condition.

The most common surgery to treat Chiari malformation is posterior fossa decompression. It creates more space for the cerebellum and relieves pressure on the spinal cord. The surgery involves making an incision at the back of the head and removing a small portion of the bone at the bottom of the skull (craniectomy). In some cases the arched, bony roof of the spinal canal, called the lamina, may also be removed (spinal laminectomy). The surgery should help restore the normal flow of CSF, and in some cases it may be enough to relieve symptoms.

Next, the surgeon may make an incision in the dura, the protective covering of the brain and spinal cord. Some surgeons perform a Doppler ultrasound test during surgery to determine if opening the dura is even necessary. If the brain and spinal cord area is still crowded, the surgeon may use a procedure called electrocautery to remove the cerebellar tonsils, allowing for more free space. These tonsils do not have a recognized function and can be removed without causing any known neurological problems.

The final step is to sew a dura patch to expand the space around the tonsils, similar to letting out the waistband on a pair of pants. This patch can be made of artificial material or tissue harvested from another part of an individual's body.

Infants and children with myelomeningocele may require surgery to reposition the spinal cord and close the opening in the back. Findings from the National Institutes of Health (NIH) show that this surgery is most effective when it is done prenatally (while the baby is still in the womb) instead of after birth. The prenatal surgery reduces the occurrence of hydrocephalus and restores the cerebellum and brain stem to a more normal alignment.

Hydrocephalus may be treated with a shunt (tube) system that drains excess fluid and relieves pressure inside the head. A sturdy tube, surgically inserted into the head, is connected to a flexible tube placed under the

skin. These tubes drain the excess fluid into either the chest cavity or the abdomen so it can be absorbed by the body.

An alternative surgical treatment in some individuals with hydrocephalus is third ventriculostomy, a procedure that improves the flow of CSF out of the brain. A small hole is made at the bottom of the third ventricle (brain cavity) and the CSF is diverted there to relieve pressure. Similarly, in cases where surgery was not effective, doctors may open the spinal cord and insert a shunt to drain a syringomyelia or hydromyelia (increased fluid in the central canal of the spinal cord).

What research is being done?

The mission of the National Institute of Neurological Disorders and Stroke (NINDS) is to seek fundamental knowledge of the brain and nervous system and to use that knowledge to reduce the burden of neurological disease. The NINDS is a component of the National Institutes of Health (NIH), the leading supporter of biomedical research in the world. The NINDS conducts research and provides research grants to major medical research institutions across the country.

Genetics

The NIH is researching the genetic factors that increase the risk of developing Chiari malformation and related brain disorders. Recent studies have identified gene mutations in the PI3K-AKT signaling pathway that cause brain overgrowth that may accompany

hydrocephalus, CM, and other brain disorders. To better understand the genetic factors responsible for Chiari I malformation, NINDS scientists are looking for other gene mutations that could act through PI3K-AKT signaling and other pathways. These studies could lead to new diagnostic tests and better treatments options for Chiari malformations and other developmental brain disorders.

Brain mechanisms

Certain signals at the midbrain-hindbrain (MHB) boundary tell the brain to properly develop the cerebellum and other parts of the brain. However, how these brain regions are initiated, formed, and maintained is not well understood. NINDS scientists are studying zebrafish embryos in order to gain a better understanding of how the MHB forms. This will provide valuable insights into human brain development, particularly the cerebellum. Other investigators are studying the expression of different growth factors on the development of the brain, skull, spine, and spinal cord. Interference with normal gene function through gene mutation or environmental factors may influence the development of CM.

Treatments

The goal of treating syringomyelia with surgery is to remove the syrinx and prevent further spinal cord injury. Little is known about the effect of surgery over time on muscle strength, pain level, and overall function. NINDS scientists are examining individuals who either have syringomyelia or are at risk of developing the disorder, including those with CM. By recording more than 5 years of symptoms, muscle strength, general level of function, and MRI scan findings from individuals who receive standard care, researchers hope to obtain more information about factors that influence its development, progression, and relief of symptoms. Study results may allow doctors to provide more accurate recommendations to individuals with syringomyelia regarding optimal surgical or non-surgical treatments.

More information about research on Chiari malformation supported by NINDS and other NIH Institutes and Centers can be found using NIH RePORTER (projectreporter.nih.gov), a searchable database of current and past research projects supported by NIH and other federal agencies. RePORTER also includes links to publications and resources from these projects.

Where can I get more information?

For more information on neurological disorders or research programs funded by the National Institute of Neurological Disorders and Stroke, contact the Institute's Brain Resources and Information Network (BRAIN) at:

BRAIN

P.O. Box 5801
Bethesda, MD 20824
800-352-9424
www.ninds.nih.gov

Information also is available from the following organizations:

American Syringomyelia & Chiari Alliance Project

P.O. Box 1586
Longview, TX 75606-1586
903-236-7079
800-272-7282
<http://asap.org>

Chiari and Syringomyelia Foundation

29 Crest Loop
Staten Island, NY 10312
718-966-2593
www.csfinfo.org

March of Dimes Foundation

1275 Mamaroneck Avenue
White Plains, NY 10605
914-997-4488
888-663-4637
www.marchofdimes.com

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Arlington, VA 22209

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