Chiari Malformation: A Literature Review

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Abstract

This literature review was designed to address a pathology that has recently become more commonly diagnosed among individuals with the help of Magnetic Resonance Imaging. The purpose is to define and discuss information previously introduced in peer-reviewed journal articles and textbooks for a thorough understanding of Chiari Malformation. There are many types of Chiari Malformations. It is a congenital condition characterized by the protrusion of brain tissues into the spinal canal through the foramen magnum. The exact cause of the disease remains unknown. A few hypotheses have been formed and are discussed in this paper. Diagnostic imaging techniques such as Radiography, Computed Tomography, and Magnetic Resonance Imaging are used for the diagnosis of Chiari Malformations. The symptoms that may or may not be present vary among individuals. The treatment methods depend on the severity of the symptoms.
The coordination of motion and balance of an individual is controlled by the cerebellum. It is located inside the base of the skull in the cavity known as the posterior fossa. The posterior fossa houses the cerebellum, brainstem, and cranial nerves 5-12.\(^1\) The cavity behind the brainstem and in front of the cerebellum that holds cerebral spinal fluid (CSF) is known as the fourth ventricle. The cerebellum is composed of two lateral halves or hemispheres. Two small protrusions called the cerebellar tonsils are located underneath the cerebral hemispheres. All of these structures are located just above the foramen magnum which is an oval-shaped opening at the base of the skull. The spinal cord enters the skull through the foramen magnum and connects to the brainstem.

Various alterations to the posterior fossa, cerebellum and foramen magnum such as Chiari Malformations have been noted throughout research with the help of diagnostic imaging. A Chiari Malformation is characterized by an anatomic defect at the base of the skull in which the cerebellum and brain stem herniate through the foramen magnum into the cervical spinal canal (see Figure 1).\(^2\)

![Figure 1. A sagittal MRI image demonstrates the cerebellar herniation through the foramen magnum at the base of the skull circled in yellow.\(^3\)](image-url)
The material presented in this paper will give a better understanding regarding the different classifications, etiology, clinical manifestations, diagnosis and treatment for Chiari Malformations by analyzing previous research and studies. The best way to diagnose Chiari Malformation is with the help of Magnetic Resonance Imagining (MRI), so the information will include important specifics about the aspects of diagnostic imaging.

Chiari Malformation is named after a German pathologist, Professor Hans Chiari. In 1981, he classified the different malformations into types I, II, III and IV based on the findings of autopsies he was performing. Julius Arnold made additional contributions to Type II Chiari Malformation. Therefore, in his honor, Type II was later named Arnold-Chiari Malformation. Future researchers did not classify later findings of Chiari Malformation using Roman Numerals. Instead, Type 0 and 1.5 were added to Chiari’s original four.

**Classification**

Chiari Malformation Type I is the most common type generally diagnosed in adolescents or adults. It occurs during fetal development and is characterized by the downward displacement of the cerebellar tonsils beneath the foramen magnum by at least 3-5 millimeters. The displacement can block the normal CSF flow between the intracranial space and the spinal cord. Anomalies where the cervical area of the spine compresses into the base of the skull resulting in brainstem compression is noted in 30-50% of patients with Chiari Malformation Type I. Lateral curvature of the spine, known as Scoliosis, may be present. Spina Bifida is noticed in conjunction with Chiari Malformation as well. This condition is the result of incomplete development of the
spinal cord and its protective covering. Part of the spinal cord is exposed and may extend out a patient’s back in a sac-like pouch. Syringomyelia may also be present (see Figure 2). Syringomyelia is a disorder where a CSF-filled tubular cyst, also known as a syrinx, forms within the central spinal cord. As the syrinx grows it destroys the spinal cord causing some of the symptoms of Chiari Malformation.

Figure 2. A sagittal MRI image where the blue arrow represents the Chiari Malformation extending below the foramen magnum with little CSF flow. The red arrow represents the syringomyelia.

Chiari Malformation Type II becomes symptomatic in infancy or early childhood as it is more severe than Type I. It is characterized by the caudal displacement of the medulla, fourth ventricle and cerebellar tonsils into the cervical spinal canal. Chiari Malformation Type II commonly occurs in children with Spina Bifida or Myelomeningocele. Myelomeningocele is a congenital condition in which the spinal cord and column do not close properly during fetal development. Because of this, the spinal cord and the protective membrane may protrude similar to Spina Bifida. The majority of patients with Chiari Malformation Type II also have hydrocephalus, an excessive buildup of CSF in the brain. Chiari Malformations often block the flow of CSF between the skull
and spine. Therefore, pressure builds up in the brain causing an abnormally large head or mental defects. A flexible tube called a shunt may need to be placed from the head to under the skin in the chest or abdomen to divert and drain the excess fluid to be absorbed by the body.\(^6\)\(^7\)

Chiari Malformation Type III is very rare. This type is characterized by a portion of the cerebellum and brainstem pushing out through an occipital encephalocele. An encephalocele is an abnormal opening in the back of the skull (see Figure 3).\(^6\) This leads to a bony defect similar to spina bifida. The meninges or brain tissue herniates outside the skull. Chiari Malformation Type III has a high and early mortality rate or severe neurological deficits in surviving patients.

![Figure 3](image)

**Figure 3.** *This axial MRI image demonstrates an occipital encephalocele protruding out the back of the skull circled in yellow.*\(^8\)

Chiari Malformation Type IV is the most severe and rarest form with an extremely high mortality rate in infancy. The cerebellum does not develop normally during pregnancy. Other malformations of the brainstem may also be present.

Chiari Malformation Type 0 is characterized by an alteration in the cerebrospinal fluid hydrodynamics at the level of the foramen magnum not necessarily with cerebellar
tonsil herniation.\textsuperscript{2,4} Patients with this type of Chiari Malformation often develop syringomyelia. Some physicians do not consider Type 0 to be a Chiari Malformation because there is no cerebellar tonsil herniation.\textsuperscript{9}

Chiari Malformation Type 1.5 is just a more advanced form of Type I. The entire cervicomedullary junction protrudes through the foramen magnum, not just the cerebellar tonsils.\textsuperscript{10} Diagnosis of Chiari Malformation Type 1.5 are rare because some doctors are not familiar with this type.

**Etiology**

Chiari Malformations are due to the developmental failure of the brainstem, cervical region of the spine and base of the skull including the foramen magnum. The exact cause of the developmental failures leading to Chiari Malformations is unknown. Chiari Malformations affect individuals of every race and ethnicity, but some studies suggest that females are more often affected versus males. The abnormally small posterior fossa may lead to the growing brain to be pushed down into the foramen magnum. Exposure to harmful substances or not enough vitamins and nutrients during fetal development, post-natal osseous growth disturbances, or a genetic association are hypothesized causes of the the small posterior fossa.\textsuperscript{6,11,12} A number of studies suggest that genetic factors play an important role in Chiari Malformations. Research has been conducted on familial aggregation and a higher degree of concordance in monozygotic twins.\textsuperscript{12} To date, no single gene has been linked to Chiari Malformations. A study was conducted sampling 415 patients with Chiari Malformation symptoms. A diagnosis was made based on cerebellar tonsil herniation of greater than 3 mm on these participants. The control sample was composed of 524 healthy blood donors that did not have Chiari
Malformation. 58 developmental genes were tested, but more confirmatory studies are needed as this was one of the first studies of its kind.

Another study was conducted using MRI images from 23 families with 71 affected individuals. The heritability and cranial morphology correlations were measured among the individuals. A linkage of over 10,000 single-nucleotide polymorphisms (SNPs) across the genome were used to identify regions of linkage to Chiari Malformation. A SNP is the most common variation in the human genome and occurs approximately once every 100 to 300 bases. The study suggested linkage to Chromosome 9 and 15.

Based on the results of the studies, it is suggested that Chiari Malformations originates as a disorder of para-axial mesoderm. This is consistent with the formation of a small posterior fossa. It is possible to acquire Chiari Malformation instead of being born with the condition. Any circumstance in which space is occupied in the posterior fossa such as a tumor, arachnoid cyst, hematoma or hydrocephalus may cause a Chiari Malformation to develop.

Clinical Manifestations

The signs and symptoms of Chiari Malformation vary from person to person. Some individuals may be asymptomatic with Chiari Malformation being an incidental finding. The symptoms of Chiari Malformations involve various areas of the central nervous system. The symptoms have been grouped into five syndromes: increased intracranial pressure, involvement of the cranial nerves, brainstem compression, spinal cord syndrome and cerebellar syndrome (see Table 1).
Table 1. Signs and symptoms related to Adult Chiari Malformation.\textsuperscript{15}

<table>
<thead>
<tr>
<th>System</th>
<th>Subjective Symptoms</th>
<th>Objective Signs</th>
</tr>
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<tbody>
<tr>
<td>Ocular</td>
<td>Transient visual obscurations, photophobia, diplopia, retro-orbital pressure, visual field abnormality</td>
<td>Papilledema, absent venous pulsations, decreased acuity, extraocular muscle palsy</td>
</tr>
<tr>
<td>Otological</td>
<td>Dizziness, tinnitus, decrease hearing, ear pressure, vertigo, hyperacusis</td>
<td>Nystagmus sensorineural hearing loss, abnormal vestibular testing</td>
</tr>
<tr>
<td>Lower Brainstem, lower cranial nerves</td>
<td>Dysphagia, dysarthria, sleep apnea, throat pain, palpations, syncope, shortness of breath, hypertension</td>
<td>Impaired gag reflex, vocal cord paralysis, hypoglossal nerve palsy, spinal nerve palsy</td>
</tr>
<tr>
<td>Cerebellar</td>
<td>Unsteady gait, poor coordination, impaired fine motor function, tremor</td>
<td>Dysmetria, ataxia</td>
</tr>
<tr>
<td>Sensory systems</td>
<td>Retro-orbital and occipital headaches, cervical pain, facial and acral numbness, paresthesia and pain, poor position sense, burning dysesthesia</td>
<td>Analgesia, impaired proprioception</td>
</tr>
<tr>
<td>Motor systems</td>
<td>Weakness</td>
<td>Weakness, spasticity, hyperreflexia</td>
</tr>
<tr>
<td>Other</td>
<td>Chronic fatigue, altered recent memory, nausea, vomiting, incontinence, impotence, trophic disturbances</td>
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Headaches are seen in 80% of patients that are experienced occipitally and may radiate into the neck and shoulders. The symptoms may worsen with exertion such as coughing, sneezing, straining or neck extension. Syringomyelia, often in conjunction with
Chiari Malformations, is associated with a variety of symptoms. The symptoms depend on the size and location of the syrinx and include loss of muscle mass, weakness, numbness, chronic pain or scoliosis.\textsuperscript{9}

**Diagnosis**

The diagnosis of Chiari Malformations is related to the patient’s symptoms. Some patients experience symptoms specific of Chiari Malformation while others experience a gradual onset of symptoms that are often misdiagnosed with another condition. Some patients do not experience any symptoms at all. In this case the finding is incidental as the patient had neuroimaging for another reason. Adult patients often go 5 years before being properly diagnosed with Chiari Malformation.\textsuperscript{15,16}

The diagnosis of Chiari Malformations is established with diagnostic imaging. The diagnostic imaging modality of choice is MRI. In 1985, MRI became widely available in the clinical practice.\textsuperscript{2} MRI is the use of magnetic radiofrequency pulses to create 2D images. Images in the sagittal plane, which divides the body into left and right halves, allows for the determination of the cerebral tonsil herniation (see Figure 4).
Figure 4. A sagittal T1-weighted MRI image of the brain. The yellow line represents the lower limit of the posterior cranial fossa. It is the reference point for measuring the cerebellar tonsil herniation.¹⁷

MRI can also confirm compression of the posterior fossa subarachnoid spaces, overcrowding in the posterior fossa, peg-shaped tonsils, a small posterior fossa, increased slope of the tentorium, medullary kinking and basilar impression.¹⁵ Both brain and cervical spine scans should be performed to rule out any associated intracranial anomalies like hydrocephalus and syringomyelia (see Figure 5). Contrast should be utilized for the scans to help validate any abnormal pathology, but especially in the case of a syrinx. Contrast allows for visualization of the flow of CSF. It can be beneficial to investigate if the Chiari Malformation is blocking the flow of the CSF between the brain and spine.¹⁶

Figure 5. An axial MRI of a child with hydrocephalus on the left and a normal brain for comparison on the right. The ventricles are noted by the white arrows. The brain with hydrocephalus demonstrates enlarged ventricles compared to the normal brain.¹⁸

Other diagnostic imaging modalities that may be utilized include Diagnostic Radiography and Computed Tomography (CT). Both of these modalities utilize radiation to create their images. Skull and cervical spine radiographs have little diagnostic value when it comes to Chiari Malformations. Radiographs are only able to display boney
abnormalities. CT is able to obtain thin slices through the spine and skull to be reconstructed into the three planes, coronal, axial and sagittal. The thin slices can better illustrate boney abnormalities.

**Treatment**

Chiari Malformation treatment is dependent upon the individual’s symptoms. Patients with Chiari Malformation that do not experience symptoms are often not treated at all. Conservative treatments such as pain medications, massage therapy or activity reduction are usually suffice in individuals with mild symptoms. Patients with either no symptoms or mild symptoms are checked regularly using MRI to monitor the progression of the condition.

Patients that are very symptomatic are treated with surgery to ease and stabilize their symptoms. Fifty percent or less of people diagnosed with Chiari Malformation actually undergo surgery. The goal of the surgery is to stop the progression of changes in the anatomy of the brain and spinal cord. Posterior fossa decompression is the operation of choice. It is performed by neurosurgeons to create more room at the base of the skull to restore CSF flow between the brain and spinal canal. There are many steps involved. The first thing that will happen in surgery is a craniotomy, when a piece of the skull is removed. A laminectomy may be performed to remove one or more of the vertebrae. A duraplasty is when the tough outer covering of the brain is cut open and a patch is sewn in to make it bigger. The cerebellar tonsils are sometimes shrunk down by cauterizing. Mesh or an artificial plate is then inserted where the craniotomy took place (see Figure 6). A typical surgical experience includes a 4 hour surgery, one night in the ICU, 3-4 days in the hospital and rest at home for 3 weeks to 3 months.
Chiari Malformation findings are becoming more common as the use of diagnostic imaging has increased. Sagittal MRI images are used to help diagnose and classify the different types of Chiari Malformations. Researchers are working to narrow down the genetic aspects that could possibly be the cause of this condition. The symptoms of Chiari Malformations depend on which type and vary from person to person. An occipital headache is the most common symptom among patients with Chiari Malformation. Some patients do not experience any symptoms at all, while others range from mild to severe. Conservative treatments should be considered for patients with mild symptoms. Neurosurgeons use the MRI findings along with careful consideration to help determine if a patient is a candidate for surgical intervention. The benefits of surgery should always outweigh the risks. The goal of treatment is to allow adult and adolescent patients with Chiari Malformations to continue to carry out activities in their normal everyday life, or extend the life of an infant or child.

Figure 6. *This image illustrates the steps of the posterior fossa decompression surgery.*

Conclusion
AMA References

1. Aans.org. The American Association of Neurological Surgeons. 2015. Available at:


16. Labuda R. Chiari Malformation: Diagnosis. 2015.


