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**Review Article**


# Chiari Malformations: A Review of the Current Literature


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## Abstract

Chiari malformations (CM) are a group of malformations involving the posterior fossa and rhombencephalic structures that may have other associated intra- or extracranial defects. Initially, the 4 classic varieties were described by the Austrian pathologist Hans Chiari towards the end of the 19th century; years later, the Swiss pathologist Julius Arnold described a case of type II CM with associated myelomeningocele. years later, two of his students used the term "Arnold-Chiari Malformation", this term is often misused for all types.

Currently it is estimated that the incidence and prevalence data are underestimated due to the asymptomatic presentation of the pathology in many cases and the limited access to health services of different populations at a global level, for this reason the epidemiological data vary between the literature. The original classification has been modified by contributions from various authors in order to more precisely describe the variety of morphological findings; currently the classification includes CM types 0, I, 1.5, II, III and IV. The symptoms and associated pathological conditions vary for each type and type III and IV are the most severe forms. Diagnosis requires that secondary causes of cerebellar tonsillar ectopy (main finding of CM) be ruled out, along with a physical examination, neurological evaluation, and complementary tests; however, diagnosis requires resonance neuroimaging to demonstrate the findings.

The management basically consists of performing a decompression of the posterior fossa and whose main indication is to reduce the symptoms, however, it should be noted that there is no global agreement among specialists on the subject of surgical management, for which each patient must be individualized by your treating physician and the decision made is subject to individual criteria.



**Keywords:** Chiari malformation; Cerebellar tonsillar ectopy; Neurological evaluation; Surgical management; Pathological conditions.




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## Introduction

The Chiari Malformations are a group of primary pathologies of interest in medical care. This article reviews the subject based on the literature on the definition, classification, diagnosis and surgical management, detailing each type. It seeks to generate knowledge on the subject that allows its identification and approach to patients during clinical practice.

## Definition

Group of malformations of the posterior fossa and rhombencephalon (medulla oblongata, pons and cerebellum) with or without other associated intra-or extracranial defects [1].

## History

In the early 1890s, the Austrian pathologist Hans Chiari described four congenital deformities, the four classic varieties named after him. The 4 were different degrees of affectation of structures of the posterior fossa [2]. In 1894, the Swiss pathologist Julius Arnold described a case of type II Chiari malformation with associated myelomeningocele; then, in 1907, two of his students used the term "Arnold-Chiari malformation" in a report of four patients with myelomeningocele and herniation of the rhombencephalon [3].

Currently the term Arnold - Chiari is used to refer to type II Chiari malformation with associated myelomeningocele, for the rest Chiari malformation (MC) and the corresponding subtype should be used.

## Epidemiology

- Type I CM is the most frequent, it is present in approximately 0.5 to 3.5% of the general population [1].
- The data on the incidence and prevalence of Chiari malformations vary according to the literature that is reviewed.
- Most of the diagnoses of type I CM are made incidentally as a finding in neuroimaging performed for some other reason [2].
- Since the majority of patients with type I CM are asymptomatic and not all patients have access to resonance neuroimaging, it is believed that the prevalence of the disease is underestimated [2,4].
- The symptomatic presentation of CM is more common in adolescents and young adults, it is believed that it is due to the ability of this population to specify their symptoms, different from young children and infants in whom it is thought that the symptoms can present for many other causes [4].
- Early diagnosis has been increasing, due to greater access of the pediatric population to health services.

## Risk factors

- As age increases, the probability of the cerebellar tonsils reaching a position greater than 5 mm below the foramen magnum decreases, this is due to age-related changes [4].
- Most studies report a higher prevalence in females than in males, both in the adult and pediatric population; however, the ratio varies greatly. The association of syringomyelia is also greater in women than in men with CM.
- The relationship between ethnicity, race and MC varies between regions


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in which studies have been carried out to be determined [4].

## Morphological classification of chiari malformations

Originally described by Hans Chiari, in more recent years it has received contributions from different authors to describe findings with greater precision; they are classified according to the morphological findings as follows [2]:

- Chiari type 0: presence of symptoms of type I CM, with caudal displacement of the cerebellar tonsils of less than 5 mm with or without associated cervical syringomyelia [1,5].
- Chiari type I: caudal migration of one or both cerebellar tonsils at least 5 mm below the foramen magnum, measured from McRae's line [1].
- Chiari type 1.5: it is considered a progression of the type I malformation, with characteristics that place it between type I and type II, such as a greater descent of the cerebellar tonsils with some involvement of the stalk less than that present in type II, without associated spinal dysraphism [1].
- Chiari type II: caudal migration of the cerebellar vermis, cerebellar tonsils, brainstem, and fourth ventricle through the foramen magnum, associated with spinal dysraphism [1,6].
- Chiari type III: severe form of herniation of the rhombencephalon (cerebellum with or without stalk), low occipital or high cervical meningoencephalocele, with other severe neurological problems and developmental defects [1].
- Chiari type IV: Severe cerebellar hypoplasia or aplasia, should not be considered as rhombencephalon hernia, so its inclusion in the classification is discussed [1].
- Chiari type V: given the finding of agenesis of the cerebellum and descent of the occipital lobe in the posterior fossa with herniation through the foramen magnum described in the article "A new form of herniation: the Chiari V malformation", Tubbs et al. propose using the term type V MC for this extremely rare type of malformation [1,7].

## Chiari malformation type 0

Initially the term MC type 0 described patients with cervical syringomyelia and displacement of the cerebellar tonsils less than 3 mm below the foramen magnum, which improved after decompression of the posterior fossa, it was believed to be a variant of CM type I; It is currently considered another type of Chiari malformation and includes patients who present symptoms of type I CM with anomalies in the configuration of the posterior cranial fossa and displacement of the subthreshold cerebellar tonsils (less than 5 mm), even in the absence of cervical syringomyelia [5]. The etiology is multifactorial, with a predominance of the genetic component, as in type I CM [8].

## Chiari malformation type I

It is the result of multiple mechanisms that lead to the displacement of the affected nervous tissue, it has been proposed as the main one, a reduced volume of the posterior fossa, which when increasing the pressure, leads to the displacement of the cerebellar tonsils through the foramen magnum [1].


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There are other causes of tonsillar herniation that are not Chiari malformation, although the findings are similar, it has an impact on the treatment; These causes include hydrocephalus, intracranial lesions, prolonged lumboperitoneal shunt, CSF fistula, achondroplasia, etc. [9]. Most of the patients are asymptomatic; however, manifestations usually present in early adulthood. In minor patients, it is common for excessive crying and irritability to appear as the main symptoms, which are not very specific. The main symptom is occipital headache/neck pain (suboccipital), with oppressive, continuous characteristics, sometimes radiating to the retroocular region, which worsens with Valsalva maneuvers; other symptoms include nausea, dizziness, difficulty swallowing, coordination disturbances (depending on the degree of compression on posterior fossa structures). In cases associated with syringomyelia, motor and sensory disorders, alteration of reflexes, suggestive of myelopathy, may occur. The neurological examination in most cases is normal, the most frequent finding being balance disturbance of vestibular origin, with difficulty walking in tandem [9].

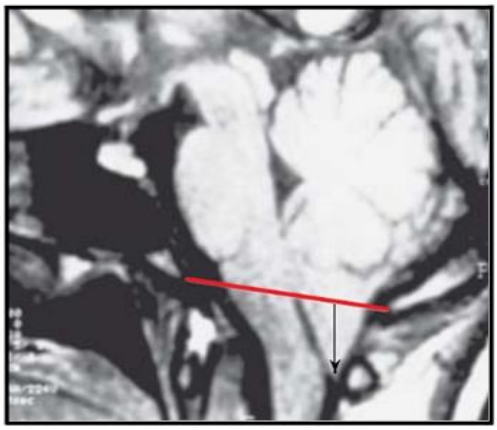

**Figure 1:** Note the descent (arrow) of the cerebellar tonsils below McRae's line (red line). CM type I.
**Image taken from:** Anatomy and pathology of the craniovertebral junction [10].

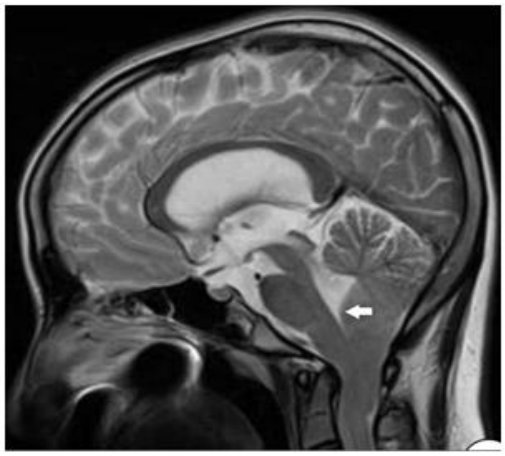

**Figure 2:** In the mid-sagittal MRI section, findings of type 1.5 MC are observed: herniation of the cerebellar tonsils of more than 5 mm and descent of the obex (arrow).
**Image taken from:** Chiari 1.5 malformation: an advanced form of Chiari I malformation [11].


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**Chiari malformation type 1.5**

It is described as an extension of the cerebellar tonsils of 5 mm or more below the foramen magnum accompanied by the brainstem (obex located below the foramen). Called type 1.5 CM because brainstem descent occurs in a similar way to type 2 CM but without the dysraphism present in these patients [12].

**Chiari malformation type II**

It is the consequence of an alteration in intrauterine neurological and skeletal development; According to McClone and Knepper, [13] the malformations present in type II CM are due to a defect in the closure of the fetal neural tube allowing cerebrospinal fluid (CSF) to escape from the entire central nervous system (CNS) through of the closure defect at the distal level, this CSF leak causes the pressure that maintains the distension of the developing ventricles to be lost, this failure in the development of the ventricular system causes inadequate neuronal growth and affects the formation of surrounding tissues, resulting in a small posterior cranial fossa, insufficient to house the cerebellum and other growing structures, this disproportion between the posterior cranial fossa and the structures it contains causes its caudal migration through the foramen magnum. In addition, the little space in the posterior cranial fossa limits the development of the spaces that will be occupied by CSF, so the flow in the ventricular system is altered, causing hydrocephalus as a consequence of the malformation [13]. Although there is usually no evidence of hydrocephalus at birth, it becomes evident in the days and weeks ahead; Babies in whom hydrocephalus worsens may present symptoms such as weak high-pitched crying, stridor, dysphagia, regurgitation, present aspiration pneumonia, in the most severe cases, apnea may occur, the most frequent cause of sudden death in these patients. They can also present upper limb motor deficits, either due to cervicomedullary compression typical of Chiari or due to the presence of syringomyelia [13].

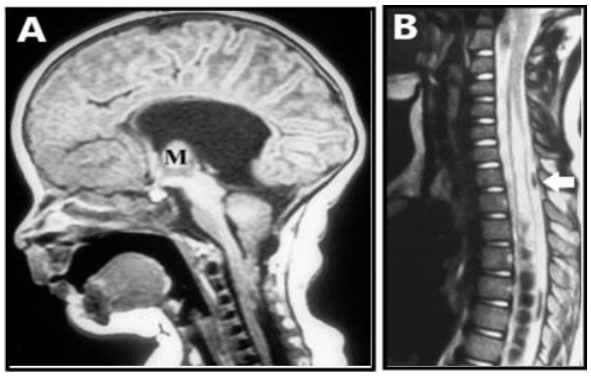


**Figure 3 A:** In the mid-sagittal section, findings of type II CM with midline lesion associated (M).3B: Syringomyelia in cervicothoracic thoracic portion (arrow).
**Image taken from:** The Chiari II malformation: cause and impact [13].

**Chiari malformation type III**

It is one of the rarest forms of CM, characterized by low occipital or high cervical encephalocele with bone defect and herniation of structures of the posterior cranial fossa through the foramen magnum. Although its etiology is still not entirely

clear, the main theory suggests a defect in neurulation during the development of the ventricular system that causes a hypoplastic posterior cranial fossa, associated with a defect in endochondral ossification; It has a higher mortality rate than types I and II. The clinical presentation is typically an occipital mass palpable from birth, which increases in size with the growth of the baby.

The severity of the clinical findings depends on the degree of brainstem herniation; sensory and motor alterations, hypotonia, nystagmus, stridor, dysphagia, respiratory pattern alterations may occur [14].

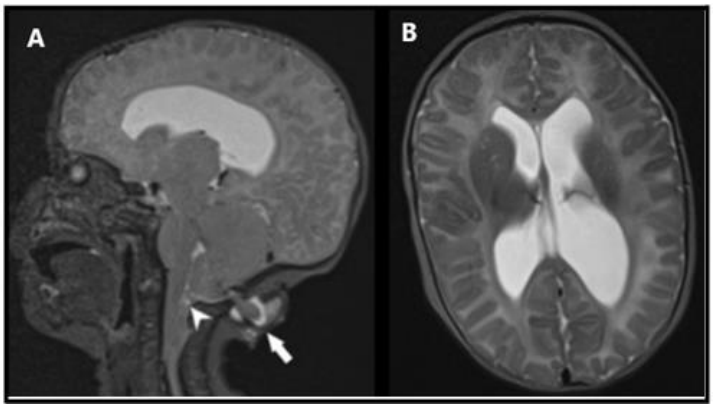


**Figure 4A:** Sagittal image of T2-weighted MRI showing suboccipital meningoencephalocele (arrow), with descent of the cerebellar tonsils (cursor). 4B: Associated hydrocephalus. Patient with type III CM.
**Image taken from:** Chiari type III malformation: Case report and review of literature [3].

### Chiari type IV malformation

This is a developmental anomaly whose origin is multifactorial, since both genetic problems and exposure to teratogens can cause this finding, which is generally incompatible with life [10]. It consists of aplasia or hypoplasia of the cerebellum, without herniation of posterior fossa structures, so its inclusion in the classification of cerebellar ectopias is debated [15].

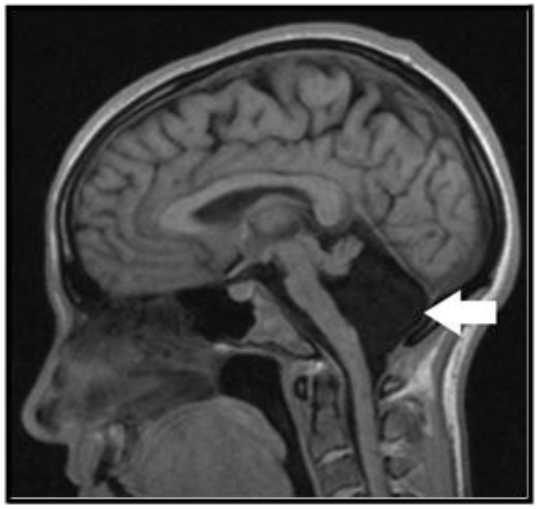

**Figure 5:** Severe cerebellar hypoplasia (arrow). Note the verticalization of tentorium.
**Image taken from:** Seram 2010 [16].


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## Diagnosis

Since symptoms are sometimes scarce or absent, it is important in patients with findings compatible with CM to rule out causes of tonsillar descent, which would not fall within the spectrum of CM but would be considered acquired or secondary tonsillar ectopia; For this, the physical and neurological examination findings are evaluated together, along with magnetic resonance imaging (simple, contrasted MRI, MRI angiography), including measurement of intracranial pressure (ICP) if it is due to intracranial hypertension [17]. By radiological definition (MRI), tonsillar herniation is defined as a descent of 5 mm or more below McRae's line or 3 to 5 mm with spike-shaped tonsils, in midsagittal section [17].

McRae's line: traced from the anterior contour of the foramen magnum (basion) to its posterior contour (opistion) (Figure 6). It is used to evaluate the descent of the cranial content through the craniocervical junction [10]. Therefore, for the diagnosis of CM, it is required to comply with the radiological criteria in patients who have ruled out another possible cause of the finding and to classify the type of CM according to the presence or absence of other findings previously described for each one. For cases of cerebellar aplasia, the term type IV MC is usually used even without ectopia of posterior fossa structures through the foramen magnum.

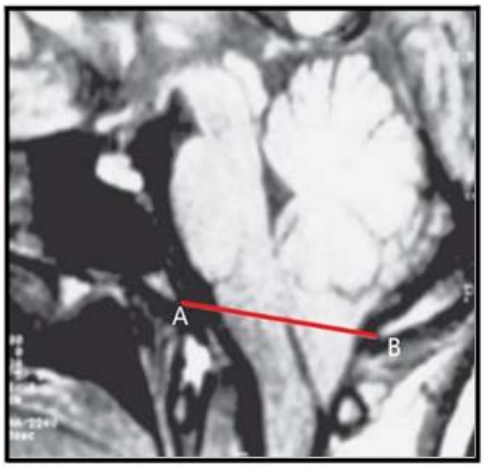


**Figure 6:** Red line: McRae's line, drawn from A: basion to B: opistion.
**Image taken from:** Anatomy and pathology of the craniovertebral junction [10].

## Management

There is still no clear global agreement on the conservative vs. surgical management of Chiari malformations. The trend is for surgical management to be carried out in symptomatic patients whose main objective is symptom improvement, clarifying that in cases of CM and the presence of other disorders such as epilepsy or cognitive disorders, surgery does not improve these conditions.

Thus, the symptoms and presence of associated syringomyelia are frequently considered the most determining criteria for surgery.

As described in the international consensus "Diagnosis and treatment of Chiari malformation type 1 in children: the


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International Consensus Document" [17], most experts agree on conservative management in asymptomatic patients, even in cases of significant tonsillar decrease (>20 mm) in NMR. The indications for surgery are:

- Asymptomatic patients with findings of type I CM with associated syringomyelia, discovered incidentally, only in cases of syrinx >5 to 8 mm or that increases in size.
- Patients with type I CM and cognitive and/or behavioral disorders who cannot clearly report their symptoms, perform clinical evaluations and tools that allow determining the presence of symptoms.

Follow-up of asymptomatic children with type I CM should be carried out with neurological evaluation and neuraxis MRI until adulthood, with a frequency that must be determined according to the clinical picture and imaging findings; Neuropediatric evaluation should be carried out in all children with symptomatic type I CM (headache and/or symptoms and signs of stalk, cerebellar or spinal cord involvement, cognitive disorders) to rule out associations [17].

In the case of adults with type I CM, the main determinants for surgical management continue to be the presence of symptoms (headache) and syringomyelia, together with findings in the neurological examination; according to the international consensus document for the diagnosis and treatment of Chiari malformation and syringomyelia in adults [18], surgery is indicated in:

- Symptomatic type I CM (disabling headache, resistant to medical management) and neurological deficit (in the evaluation).
- Type I MC with associated syringomyelia according to the radiological and clinical findings and with diagnostic tools.

Although surgery is not indicated in asymptomatic patients without syringomyelia [18], follow-up with neurological examination and neuraxis MRI is necessary, the frequency should be determined according to the findings.

**Surgical procedure**

Bone decompression of the posterior fossa is the most performed surgical management, associated with other procedures such as duroplasty and coagulation of the cerebellar tonsils.

In children [17] bone decompression alone can be performed due to its low complication rate, however there is a greater probability of requiring reoperation given the lower success rate than decompression+duroplasty, which has a higher complication rate (leakage of CSF). Therefore, decompression alone is preferred in patients without syringomyelia and decompression+duroplasty in patients with more severe symptoms and associated syringomyelia, considering the greater risk of complications; however, in clinical practice, management is more subject to the experience and opinion of the treating neurosurgeon. In adults [18], due to the risk of recurrence of symptoms, bone decompression+duroplasty is indicated in patients without associated syringomyelia and patients with associated syringomyelia, regardless of symptoms. Regarding the extent of decompression, the key region to decompress is the one in relation to the medulla oblongata and the cervical cord,


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which includes the posterior arch of $C_1$ and the foramen magnum (posteriorly delimited by the occipital bone scale). Although surgery must be planned on an individual basis for each patient, the risk of instability of the craniocervical junction is greater when laminectomy is performed up to $C_2$, so it should be avoided. In the case of duroplasty, the integrity of the arachnoid must be maintained to reduce the risk of CSF leakage, except in cases of arachnoiditis. With regard to coagulation of the cerebellar tonsils, it should be considered in patients with severe tonsillar descent (below $C_2$ level), recurrent syringomyelia, and/or severe cerebellar dysfunction, because this procedure increases morbidity.

The treatment of type II Chiari malformation requires a comprehensive evaluation of the patient's findings, type II CM is usually present in patients with spinal dysraphism, with associated hydrocephalus and symptoms that present from the moment of birth and worsen with age. As the days pass, due to the above, emergency surgery is indicated in neonates. Since in older patients the symptoms remain stable, surgery is not indicated as urgent.

Although there is no consensus on the management of type II BC, the main determinant for decision-making in the neurosurgical management of patients has been the association of hydrocephalus and the presence of symptoms that put patients' lives at risk; Given the risk of deterioration, the first step is usually to ensure CSF drainage through external ventriculostomy, which improves symptoms in most patients and decreases the morbidity and mortality rate in patients undergoing decompressive surgery of the posterior fossa (craniectomy with or without duroplasty) as definitive management for type II CM and closure of the spinal defect, with subsequent guarantee of CSF drainage by definitive shunting of the ventricular system [19].

Some authors suggest resecting the nervous tissue in encephalocele trying to preserve neurological function as much as possible and repairing the closure defect through dural and soft tissue repair [20]. Additionally, patients require management of other possible associations such as hydrocephalus (more frequent). The prognosis of each patient is subject to the neurological status prior to the intervention, other associated disorders and the amount of tissue resected in surgery [21].

## Conclusion

Chiari malformation is a term used to describe a diverse set of findings originally described by pathologist Hans Chiari in the 19th century, including abnormalities of the posterior cranial fossa and its contents. The current classification includes two more types (0 and 1.5) with clear differences between each one. Its diagnosis requires a detailed clinical evaluation, especially in search of signs and symptoms of neurological alteration and resonance imaging study. Treatment differs between each type and additionally requires intervention in other disorders that may be associated; Although there is no global expert consensus regarding surgical management, the options that exist for the management of a variety of patients have been precisely described.

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