Adult Chiari Malformation

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Learning Objectives: After reading this article, the participant should be able to:
1. Identify the epidemiologic and pathophysiologic aspects of adult Chiari malformation.
2. Describe the clinical presentation and diagnostic workup of patients with adult Chiari malformation.
3. Explain the indications for and complications of surgical treatment of adult Chiari malformation.

Interest in the adult Chiari malformation (ACM) has increased since the early 1990s. The widespread use of magnetic resonance imaging (MRI) has led to frequent detection of tonsillar ectopia, and the broad array of clinical manifestations of ACM is increasingly recognized. However, the syndrome continues to be under- or misdiagnosed, with an average delay in diagnosis of 5 years after onset of symptoms, and more than half of patients with ACM labeled psychogenic.

Definition

Most scholars agree that ACM consists of ectopic cerebellar tonsils diagnosed in adult patients. The amount of cerebellar ectopia required for the diagnosis of ACM is discussed later in this article, in the section entitled Diagnostic Imaging Modalities.

Background

In 1891, while studying the effects of hydrocephalus on the cerebellum, Chiari described various alterations that became known as the Chiari malformations (Table 1). Type I consisted of elongation of the cerebellar tonsils into cone-shaped projections into the spinal canal. Chiari's original patients had accompanying hydrocephalus, and this finding might have represented tonsillar herniation caused by increased intracranial pressure (ICP) secondary to hydrocephalus. Very few cases

were reported over the following few decades, especially in the English literature. The first description of an adult case of Chiari malformation without accompanying hydrocephalus was published by Aring in 1938. Scattered cases of ACM were reported afterward, mostly in relation to syringomyelia. It was only in the 1960s and 1970s that larger series of ACMs were published, allowing a better understanding of this entity.

Epidemiology

The advent of MRI, with its capacity for sagittal views, has revolutionized the diagnosis of ACM. The incidence of ACM seen on MRI is between 0.56% and 0.77%, and 15% to 30% of those patients are asymptomatic. Tonsillar ectopia also is identified in 0.62% of patients in brain dissections. It is estimated that more than 3500 decompressions are performed annually

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Table 1: Classification of Chiari Malformations

Malformation Type	Description
Type I	Elongation of the tonsils and the medial parts of the inferior lobes of the cerebellum into cone-shaped projections, which accompany the medulla oblongata into the spinal canal
Type II	Displacement of the parts of the inferior vermis, pons, and medulla oblongata together with elongation of the fourth ventricle (most cases are associated with spina bifida)
Type III	The entire cerebellum herniates into the cervical canal
Type IV	Cerebellar hypoplasia

Category: Spine

Key Words: Adult Chiari malformation, Clinical Presentation, Surgical Indications, Tonsillar ectopia

in the United States for Chiari malformation. The disease is most common in women, with a female-to-male ratio of 3:1. The age of onset is about 25 years (± 15 years), with the diagnosis made, on average, 5 years from onset.

There is a strong association between the Chiari malformations and syringomyelia. Between 30% and 50% of type I Chiari malformation and 45% to 90% of type II Chiari malformations have an associated syrinx, and up to 70% of syringomyelias are related to a hindbrain disorder. The syrinx associated with the ACM usually is cervical or cervicothoracic.

Etiopathology

Adult Chiari malformation results from a craniocephalic disproportion, leading to tonsillar ectopia. The primary event is a mismatch between the container (skull) and the contents (brain). A small posterior fossa is a common finding in patients with ACM. There have been reports of acquired tonsillar ectopia in patients who have a thickened calvarium and craniosynostosis, both of which decrease the intracranial volume. Tonsillar ectopia also can occur with conditions that increase the intracranial contents, mainly hydrocephalus and intracranial mass lesions (both supra- and infratentorial).

Clinical Manifestations

Physiopathology

Different mechanisms provoke the clinical manifestations of ACM. They are related to direct compression of nervous tissue or the valve effect caused at the foramen magnum by the protruding tonsils.

Direct compression of the lower brainstem and upper cord by the ectopic tonsils leads to lower brainstem and cerebellar signs. The partial valve-and-piston effect leads to intermittent obstruction to the flow of cerebrospinal fluid (CSF) between the cranial and spinal compartments; intracranial hypertension and syringomyelia are two of the consequences. With every systole, the cerebral blood volume increases, leading to brain engorgement. In normal individuals, this increase in brain volume is accommodated by the flow of CSF from the intracranial to the intraspinal compartments. In patients with ACM, however, the ectopic tonsils block this flow; instead, the tonsils are thrust caudally by the increased brain volume during systole. This caudal movement acts like a piston on the subarachnoid space around the spinal cord. The perispinal cord pressure increases, pushing the CSF into the perivascular Virchow-Robin spaces of the spinal cord and leading eventually to syringomyelia.

The obstruction of CSF flow by the partial valve effect and the displacement of CSF from the compressed spaces of the posterior fossa lead to a reduced dampening effect to pressure changes, altered CSF–brain compliance, and increased ICP. This explains the wide constellation of symptoms in the Chiari malformation, some of which do not fit with lower brainstem compression or syringomyelia but resemble some of the symptoms found in idiopathic intracranial hypertension, e.g., suboccipital headaches, episodes of retro-orbital pain and visual phenomena, Ménière's disease-like symptoms, and symptomatic worsening with exertion and Valsalva maneuver, as well as others.

Symptoms and Syndromes

The clinical manifestations of ACM involve various areas of the central nervous system, including the visual system, the neuro-otological system, the lower cranial nerves, the cerebellum and its pathways, the motor pathways, and the sensory pathways, as well as other systems. These symptoms have been grouped into syndromes. Five syndromes were recognized by Adams et al. in 1941: increased ICP syndrome; involvement of several of the cranial nerves; brainstem compression syndrome; spinal cord syndrome; and cerebellar syndrome.

There are limitations to classification by syndrome. Although some patients have one particular syndrome, in others the clinical picture does not clearly fit any one syndrome, with significant overlap between two or more syndromes. We prefer to describe the different symptoms encountered as they relate to different neurological systems,

excluding the cases with syringomyelia, which will be discussed separately (Table 2).

Table 2: Symptoms and Signs of Adult Chiari Malformation (excluding those related to syringomyelia)

System	Subjective Symptoms	Objective Signs
Ocular	Transient visual obscurations, photophobia, diplopia, retro-orbital pressure, visual field abnormality	Papilledema, absent venous pulsations, decreased acuity, extraocular muscle palsy
Otological	Dizziness, tinnitus, decreased hearing, ear pressure, vertigo, hyperacusis	Nystagmus, sensorineural hearing loss, abnormal vestibular testing
Lower brainstem, lower cranial nerves	Dysphagia, dysarthria, sleep apnea, throat pain, palpitations, syncope, shortness of breath, hypertension	Impaired gag reflex, vocal cord paralysis, hypoglossal nerve palsy, spinal nerve palsy
Cerebellar	Unsteady gait, poor coordination, impaired fine motor function, tremor	Dysmetria, ataxia
Sensory systems	Retro-orbital and occipital headaches, cervical pain, facial and acral numbness, paresthesias and pain, poor position sense, burning dysesthesia	Analgesia, impaired proprioception
Motor systems	Weakness	Weakness, spasticity, hyperreflexia
Other	Chronic fatigue, altered recent memory, nausea, vomiting, incontinence, impotence, trophic disturbances	

Table 2 presents most of the symptoms encountered in ACM. The various symptoms occur with variable frequency, and in variable combinations, which explains the particularly challenging clinical diagnosis. There are, however, some clinical features that are typical of the ACM, including worsening of symptoms with exertion, the Valsalva maneuver, coughing, straining, head dependency, and neck extension, and worsening of symptoms during the premenstrual phase. This is especially true of the headaches that are seen in 80% of patients. They are occipital and pressure-like, and they radiate to the retro-orbital area anteriorly and the neck and shoulders inferiorly. Ocular and neuro-otological disturbances occur in about 75% of patients, and cerebellar and lower brainstem disturbances occur in 50% of patients. Long tract motor and sensory alterations occur in more than 50% of patients, even when syringomyelia is not present.

The symptoms of syringomyelia are a combination of those due to the hindbrain anomaly and those due to the destruction of the spinal cord. There are two categories of symptoms caused by destruction of the spinal cord:

1. A suspended syndrome caused by the destruction of the central gray matter, leading to dissociated sensory loss (thermo-algesic sensory loss) and burning sensation in a few dermatomes. This is followed by destruction of the anterior horn motor cells, leading to a lower motor neuron syndrome, with subsequent weakness and amyotrophy.

2. A long tract syndrome, with involvement of the corticospinal tracts, leading to spasticity, or of the posterior columns, with proprioceptive sensory loss.

Adult Chiari Malformation and Trauma

The relationship between ACM and trauma is not well defined. About one of four patients with ACM cites trauma as the precipitating factor for his or her symptoms. There have been scattered reports of sudden death and spinal cord injury after trauma in patients with tonsillar ectopia. It is also known that hyperextension of the neck worsens the symptoms of ACM. Although it seems prudent for patients with symptomatic ACMs to avoid neck trauma, there are not yet enough reports to support firm recommendations concerning engagement in athletic activities for these patients.

Diagnostic Imaging Modalities

The diagnosis of ACM often is delayed. The interval from clinical presentation to diagnosis usually is about 5 years. This delay in diagnosis results, in part, from the vast array of symptoms found in patients with ACM. The diagnostic imaging modality of choice is MRI.

Magnetic Resonance Imaging

Various MRI criteria have been used to diagnose ACM, most of which address the amount of tonsillar herniation into the cervical spinal canal (Fig.1). The availability of sagittal views allows exact determination of the tonsillar ectopia. Many authors, even before the MRI era, attempted to define what constitutes an abnormal tonsillar descent, with 3 to 5 mm chosen by most radiologists as the threshold value. Mikulis et al. found that the tonsils ascend with age and suggested 6 mm as diagnostic in the first decade of life, 5 mm in the second to third decades, 4 mm in the fourth to eighth decades, and 3 mm in the ninth decade. These thresholds are not absolute: some asymptomatic patients harbor significant tonsillar ectopia, and some symptomatic patients have minimal ectopia. Ten percent of patients with ACM in Milhorat's series had ectopia of less than 5 mm, and 43% of those had syringomyelia. Meadows found a significant percentage of asymptomatic patients (14%) with ectopia of more than 5 mm. The range of 3 to 5 mm seems a relatively sensitive but not an absolute value. When treating patients with borderline ectopia, the clinician should be extremely careful before making the diagnosis of ACM unless there is other strong compelling clinical evidence. A clinicoradiological correlation remains the cornerstone of diagnosis.

Other MRI findings encountered in ACMs, to varying degrees, include: 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.
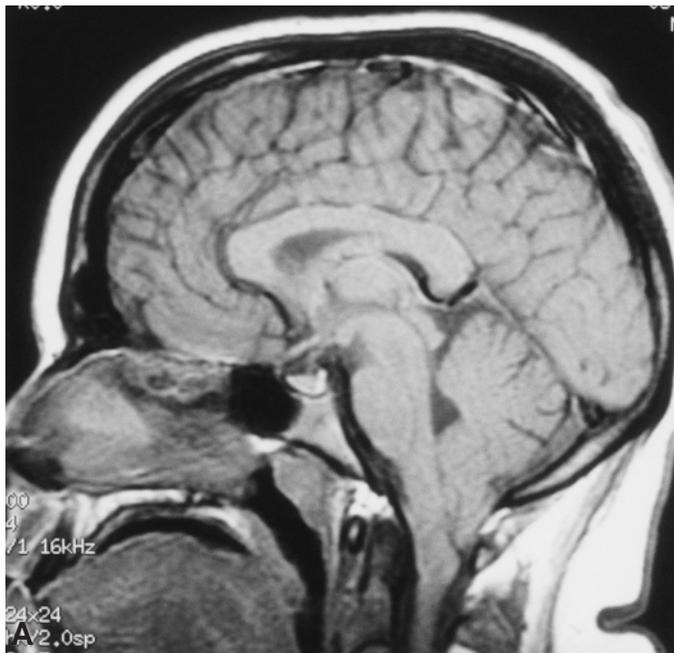
Figure 1. Preoperative *(A)* and postoperative *(B)* sagittal MR images of a patient with ACM. Preoperatively, the patient reported headaches and orthostatic visual obscurations, indicative of papilledema. Postoperatively, formation of a neo-cistern behind the tonsils is observed. The tonsils are rounder in shape.

The work-up should include brain and cervical MR scans to rule out associated intracranial anomalies (e.g., tumors, hydrocephalus) that may lead to tonsillar ectopia, as well as associated syringomyelia (Fig. 2). If syringomyelia is present, contrast-enhanced MRI is performed to rule out an associated spinal cord tumor.

MRI Cine-Flow

The failure of MRI to provide strict criteria for tonsillar ectopia led to the investigation of a potential diagnostic value for cine-flow MRI. Some of the findings with this modality were caudal systolic motion of the tonsils and loss of flow behind the tonsils or anterior to the brainstem. So far, cine-flow MRI has not been proven to be of major diagnostic or prognostic value.

Radiography

Cervical and skull radiographs are of limited diagnostic value. They usually reveal associated bony anomalies, which may be present in 30% to 50% of patients with ACMs. These anomalies include basilar invagination, atlanto-occipital fusion, atlanto-axial assimilation, the Klippel-Feil deformity, congenital spinal stenosis, hemivertebrae, and os odontoideum, among others. The delineation of potential osseous anomalies on radiographs is useful during surgical intervention for ACMs.

Computed Tomography

Computed tomography is useful primarily in obtaining thin-section bone windows to delineate any associated bony anomalies.

CT Myelography

Magnetic resonance imaging has largely replaced CT myelography as a diagnostic tool. A CT myelogram can delineate the ectopic tonsils and the compression of the posterior fossa cisterns.

Differential Diagnosis

The widespread use of MRI has made it possible to diagnose an increasing number of cases of tonsillar ectopia. Some patients are asymptomatic, whereas others have symptoms that are not specific for ACMs. This is a double-edged sword: patients who do have an ACM may be missed and labeled as having a variety of other diseases, but, on the other hand, patients with borderline ectopia may undergo decompression for ACMs when their symptoms actually are caused by other pathologies. The diagnosis of ACM should be one of exclusion. Clinicians must be careful in excluding other entities with a similar presentation before embarking on surgical decompression. Some of the entities to be entertained in the differential diagnosis are as follows:

- *Multiple sclerosis.* The diagnoses of multiple sclerosis and ACM often are confused, because the two conditions have similar age and sex distributions, and both involve multiple neurological functional systems.

- *Fibromyalgia and chronic fatigue syndrome (CFS).* Fifty-seven percent of patients with ACMs report chronic fatigue, and 39% have recent memory loss (Milhorat et al.), statistics that explain the frequent mislabeling of patients with ACMs as suffering from fibromyalgia or CFS. However, patients with CFS or fibromyalgia do not necessarily have ACM. Physicians must be very careful in evaluating patients diagnosed with chronic fatigue syndrome or fibromyalgia.

- *Psychogenic disorder.* The wide array of symptoms reported by some patients with ACMs does not fit within a specific syndrome or a single topography and may resemble some psychiatric entities. There is a tendency, therefore, for clinicians to dismiss some of these patients' symptoms as psychogenic. Patients with ACMs may have secondary alterations in their affect due to the chronic nature of their symptoms, their seemingly noncoherent nature, and the skeptical attitude of their families, friends, and physicians.

- *Migraine disorder.* The headaches experienced by patients with an ACM often are mislabeled as migraine headaches. A careful history, however, can delineate the type of headache.

- *Idiopathic intracranial hypertension (IIH).* There is an overlap between patients with ACMs and those with IIH. Six percent of patients with IIH have tonsillar ectopia consistent with ACM. Experimental models of pseudo-tumor cerebri induced by jugular ligation have demonstrated secondary tonsillar ectopia. As we mentioned previously, an increase in the volume of intracranial contents, such as is seen in cerebral edema, may lead to tonsillar ectopia. Some patients with ACMs have papilledema and increased ICP. The symptoms of IIH and ACM are similar, as was discussed in the Physiopathology section. We have encountered numerous patients in our practice with IIH and associated ACM in whom initial decompression fails. The symptoms eventually resolved after treatment directed at the IIH, with either repeat lumbar puncturek, acetazolamide, or shunting.

- *Spinal cord tumors.* Because spinal cord tumors can be associated with syringomyelia and may have symptoms similar to those of syringomyelia, it is necessary to obtain contrast-enhanced MR scans to rule out associated spinal cord tumors, especially in patients with borderline ectopia.

Recommended Therapy: Indications

Treatment of ACM is mostly surgical. However, not every tonsillar ectopia, Chiari-related syringomyelia, or symptomatic ACM needs to be treated. The following factors are taken into consideration when addressing the risk/benefit ratio:

- The severity and nature of the symptoms;

- The alteration in the patient's quality of life secondary to these symptoms;

- The likelihood that these symptoms are related to the tonsillar ectopia, taking into consideration the clinical presentation, associated medical contitions, and the radiological findings;

- Associated psychological factors;

- The presence of symptomatic syringomyelia;

- The surgical complication rate; and

- The long-term results for surgical treatment of the particular symptom that is being addressed, with most series reporting a long-term success rate between 50% and 85%.

The surgical decision is highly individualized, especially because most symptoms are subjective. The expected improvement in quality of life should outweigh the potential uncertainty regarding the causal relationship between the tonsillar ectopia and the particular symptoms, the risk of the surgery, and the long-term failure rate. We reserve surgery for patients with disabling or unbearable symptoms that are likely to be related to the ACM. Some patients with minor symptoms need only reassurance that their symptoms are caused by a real disease, and that they are not dangerous or life-threatening.

In ACM associated with syringomyelia, there is concern that the destruction of spinal cord tissue may lead to irreversible neural damage. Some recent evidence has suggested conservative management for asymptomatic cases: eight of nine patients in the Nishizawa et al. series remained asymptomatic after more than 10 years' follow-up. Surgery is indicated in cases of symptomatic syringomyelia, especially when there is clinical deterioration or the patient experiences unbearable symptoms.

In patients with ACMs whose symptoms are caused by basilar invagination or compression from a ventral pannus, management follows that of the ventral pathology. This variant is not discussed in this article.

Surgical Treatment

Because craniocephalic disproportion and alteration of CSF flow dynamics between the cranial and spinal compartments at the level of the foramen magnum are the main culprits, surgery is directed at reestablishing normal flow by enlarging the foramen magnum, and, for some surgeons, enlarging the posterior fossa. A decompressive suboccipital craniectomy has been the mainstay of treatment, with numerous modifications, each of which has strong advocates. As discussed previously, patients with anterior compression are managed differently.

Surgical Techniques

Our preferred technique includes suboccipital craniectomy, C1 laminectomy, dural opening, and duraplasty. Intra-arachnoid exploration is reserved for cases in which macroscopic assessment of restoration of the CSF flow behind the tonsils and lower brainstem is not possible. A C2 laminectomy is performed in cases of tonsillar herniation below the midpoint of the C1-C2 interspace. Other variations include incising the outer leaf of the dura and leaving the inner leaf intact, intra-arachnoid exploration, tonsillar shrinkage, and fourth ventricular shunting. A variety of materials are used for the duraplasty, including pericranial and fascia lata autografts; pericardial, fascia lata, and dural allografts; bovine pericardium; and synthetic patches.

Surgical Complications

The following paragraphs present some of the typical complications of suboccipital decompression for ACM.

Cerebrospinal fluid leak. Cerebrospinal fluid leakage can be fatal if it leads to meningitis. This complication is prevented by watertight dural closure and tight fascial and skin closure. It is treated by over-sawing the incision and lumbar drainage
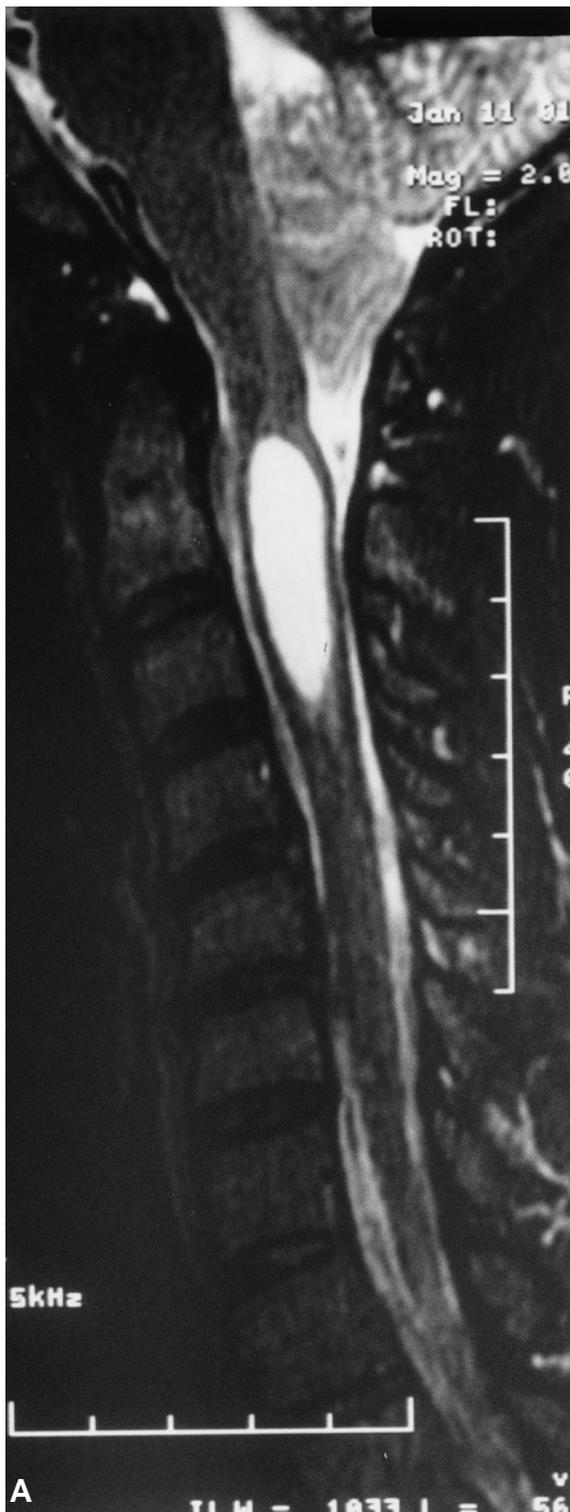
Figure 2. Sagittal preoperative *(A)* and postoperative *(B)* cervical MR image of a patient with ACM-associated syringomyelia. The patient experienced upper extremity paresthesias after a motor vehicle accident. She underwent a suboccipital decompression, after which her symptoms resolved and the syrinx disappeared.

if the leak persists. The possibility of an intracranial mass should be excluded before the drain is placed.

Pseudomeningocele. Some failures result from pseudomeningocele. Prevention is by watertight dural closure, and initial treatment is by lumbar drainage. If the latter fails, re-exploration may be indicated.

Brainstem infarction. Brainstem infarction may occur secondary to injury to the vertebral artery or posteroinferior cerebellar artery (PICA). The vertebral artery may be injured during dissection of the posterior arch of C1. The PICA can have an extradural origin, making it subject to injury during extradural exposure. It also may be injured during intradural dissection. Avoiding unnecessarily wide or aggressive dissection over the arch of C1 is advocated as a preventive measure. We avoid aggressive intra-arachnoid dissection to reduce the risk of vascular injury to the PICA. When a vascular injury occurs, arterial repair should be attempted if feasible.

Cerebellar sag. Cerebellar sag has been reported to occur following generous suboccipital decompression. It leads to intractable headaches and recurrent syringomyelia. Treatment is by suboccipital cranioplasty or ventriculoperitoneal shunting.

Results

The success rate of surgical treatment is variable and decreases with longer follow-up periods. The long-term success rate is difficult to compare between series, because the definitions of failures or recurrences tend to vary. The recurrence may be milder and more tolerable than the original condition. The long-term success rate is between 50% and 85%, with the best results obtained for headache or neck pain, followed by cerebellar symptoms. The worst results are seen with lower cranial nerve dysfunction and syringomyelia. Dense arachnoiditis also is associated with a poor outcome.

Summary

Adult Chiari malformation is diagnosed increasingly as use of MRI becomes widespread. The syndrome has a wide array of clinical manifestations, leading to frequent delay in diagnosis or misdiagnosis. Some patients with radiologically significant tonsillar ectopia are asymptomatic, whereas other patients have other diseases that mimic the clinical presentation of ACM. Treatment is indicated if patients experience significant alteration in their quality of life. The treatment is surgical, consisting of suboccipital decompression with numerous technical variations, and it has a long-term success rate between 50% and 85%.

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