Chiari malformation

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Introduction

This article includes discussion of Chiari malformation, Arnold-Chiari deformity, and Arnold-Chiari malformation. The foregoing terms may include synonyms, similar disorders, variations in usage, and abbreviations.

Overview

Chiari malformation describes a group of structural defects of the cerebellum, characterized by brain tissue protruding into the spinal canal. Chiari malformations are often associated with myelomeningocele, hydrocephalus, syringomyelia, and tethered cord syndrome. Although studies of etiology are few, an increasing number of specific genetic syndromes are found to be associated with Chiari malformations. Management primarily targets supportive care and neurosurgical intervention when necessary. Renewed effort to address current deficits in Chiari research involves work groups targeted at pathophysiology, symptoms and diagnosis, engineering and imaging analysis, treatment, pediatric issues, and related conditions. In this article, the author discusses the many aspects of diagnosis and management of Chiari malformation.

Key points

- Chiari malformation describes a group of structural defects of the cerebellum, characterized by brain tissue protruding into the spinal canal.
- Chiari malformations are often associated with myelomeningocele, hydrocephalus, syringomyelia, and tethered cord syndrome.
- Although studies of etiology are few, an increasing number of specific genetic syndromes are found to be associated with Chiari malformations.
- Management primarily targets supportive care and neurosurgical intervention when necessary.
- Renewed effort to address current deficits in Chiari research involves work groups targeted at pathophysiology, symptoms and diagnosis, engineering and imaging analysis, treatment, pediatric issues, and related conditions.

Historical note and terminology

The Chiari malformation, also known as the Arnold-Chiari malformation (or deformity), was first described by Cleland in 1883 in a child with spina bifida, hydrocephalus, and alterations of the cerebellum and brainstem (Cleland 1883; Schijman 2004). In 1891, Austrian pathologist Hans Chiari wrote an article titled “Concerning alterations in the cerebellum resulting from hydrocephalus.” He described elongation of the cerebellar tonsils and medulla below the plane of the foramen magnum in a 17-year-old woman, now referred to as a type 2 malformation. Chiari's subsequent studies expanded the spectrum of malformations in a classification system consisting of types 1, 2, 3, and later 4. In 1907, Arnold was credited with publishing 4 additional cases, and his students Schwalbe and Gredig added his name to the nomenclature. Many authors consider Chiari's contribution to be primary and, therefore, suggest that this condition be designated as Chiari malformation, without including Arnold's name (Koehler 1991).

Chiari type 1 refers to herniation of the cerebellar tonsils alone, and radiologically as simple tonsillar herniation 5 mm or greater below the foramen magnum (Elster and Chen 1992); type 2 refers to herniation of both the cerebellum and lower brainstem; type 3 refers to a rare type of brainstem herniation in association with a cervical or occipital encephalocele; type 4 involves extreme cerebellar hypoplasia and caudal displacement of the posterior fossa contents. Two additional types of Chiari malformation have been described. Chiari type 0 is defined as syringohydromyelia with distortion of contents in posterior fossa but without cerebellar tonsillar herniation (Iskandar et al 1998; Tubbs et al 2004b). Chiari type 1.5 has been characterized as caudal migration of the brainstem and cerebellar tonsils often associated with syringomyelia but without spina bifida (Schijman 2004; Tubbs et al 2004b).

Clinical manifestations

Presentation and course

Clinical presentations resulting from Chiari malformation vary along a spectrum of severity and depend on anatomic involvement. One may approach clinical presentations by their underlying pathology or pathophysiology. Another approach involves identifying cases by the presence or absence of myelomeningocele. Many individuals with Chiari 1 malformation do not have symptoms and go undiagnosed until adolescence. Alternatively, because of brainstem involvement, type 2 malformations are typically diagnosed earlier in childhood. Therefore, some categorize type 1 malformations as “adult type” and type 2 malformations as “congenital type.” Headache and neck pain are the most common presenting symptoms of Chiari 1 malformations (Aitken et al 2009). Chiari 1 malformation may present with seizures and developmental delay in children (Brill et al 1997).

Chiari malformation without myelomeningocele. Symptomatic Chiari malformation can present at any age. Children and adults present with similar symptoms and signs (Dure et al 1989; Dyste et al 1989). Saez and colleagues grouped presentations into 6 categories: (1) foramen magnum compression, (2) central cord syndrome, (3) cerebellar dysfunction, (4) bulbar palsy, (5) paroxysmal intracranial hypertension, and (6) spasticity (Saez et al 1976). Pillay and colleagues presented a simplified system, classifying patients as type A (with concomitant syrinx and central cord symptoms) or type B (with significant brainstem herniation and signs and symptoms of brain stem and cerebellar compression) (Pillay et al 1991). In their study of 35 patients, Pillay and colleagues found the frequency of clinical presentations to be the following: headache and neck pain, 73%; sensory dysesthesias or numbness, 56%; gait problems, 43%; upper extremity weakness, 25%; cranial nerve dysfunction, 23%; blurred vision, 17%; and lower extremity weakness, 15%. This system does not account for the important category of intermittent symptoms most often associated with paroxysmal intracranial hypertension. A system that adds the group with intermittent presentations to the two in Pillay's system is probably an adequate classification.

Some authors have stressed disorders of oculomotor motility, particularly downbeat nystagmus, as being important in making the diagnosis of a symptomatic Chiari malformation. Indeed, a variety of eye movement disturbances has been identified as a feature of this condition, including convergence nystagmus, internuclear ophthalmoplegia (Chiari malformation type 2), oscillopsia, horizontal nystagmus, and ophthalmoplegia.

Although eye movement disturbances may be seen, other signs are usually more prominent on presentation as the above-described classification systems attest. Particular attention should be paid to the following:

Presentations due to an accompanying syrinx. Syringomyelia, when associated with a Chiari 1 malformation, usually involves the cervical spinal cord, with accompanying symptoms or signs of upper extremity involvement. Cases involving thoracic syrinx can cause scoliosis without upper extremity findings. Idiopathic scoliosis in children can be the only manifestation of Chiari malformation with syrinx or hydromyelia.

Progressive brainstem or cerebellar dysfunction. This includes sleep apnea in adults, dysphagia, progressive myelopathy, sensorineural hearing loss, and vertigo if accompanied by neck pain or weakness, as well as other otolaryngological symptoms, disturbances of respiratory drive (Ondine curse and pneumonia with respiratory failure), and trigeminal neuralgia. Cervical pain may be a complaint.

Paroxysmal symptoms. Headache, autonomic dysfunction, syncope, and pain can be a presenting complaint of Chiari malformation, or can accompany the more classic presentations noted above. These intermittent symptoms can be cough-induced.

The nature of head pain or headaches in Chiari malformation varies and can be exertional, with characteristics of a spinal headache, either migrainous or cough induced (Pascual et al 1992; Stovner 1993). In rare cases, glossopharyngeal neuralgia may be a presenting sign of Chiari 1 malformation (Li et al 2012). Chiari I malformation has been implicated as a cause of such syndrome complexes as chronic fatigue syndrome and neurally mediated orthostatic hypotension. New techniques for early identification of Chiari malformation are being proposed, including decreased ratio of downward to horizontal smooth pursuit eye movement velocity (Kobayashi and Sugiyama 2012).

The presence and degree of hydrocephalus and ventriculomegaly. Chiari malformation can be accompanied by hydrocephalus, which is often compensated for, without signs or symptoms of, increased intracranial pressure.

Hydrocephalus may also be of the intermittent obstructive type.

Chiari malformation in those with myelomeningocele. The Chiari malformation is a common problem in patients with spina bifida, particularly myelomeningocele, where it occurs in more than 90% of individuals. Clinical manifestations of Chiari malformation in children and adults with myelomeningocele can be similar to those discussed above. The presence and severity of hydrocephalus and ventriculomegaly are particularly important clinical matters in such cases. Added to this is the infantile presentation of Chiari malformation seen in 10% to 20% of patients with myelomeningocele. Because of its severity, along with the potentially poor prognosis, the infant brainstem syndrome of myelomeningocele is considered separately. A new form of herniation of the occipital lobe through the foramen magnum is rare and has been proposed as Chiari type 5 (Tubbs et al 2012).

For children, adolescents, and adults with myelomeningocele, clinical manifestations are similar to those without myelomeningocele. Sleep apnea appears to be common in this group, both the central and obstructive types (Cochrane et al 1990). Some cases of obstructive sleep apnea appear to be related to abnormal control of pharyngeal airway patency in sleep (Waters et al 1998). Symptoms from accompanying syrinx or hydromyelia are also common, especially those of progressive upper extremity dysfunction, scoliosis, or both.

Symptoms of brainstem dysfunction such as stridor, apnea, and dysphagia, are seen in 10% to 20% of newborns and infants with myelomeningocele, and can be ominous signs. In some series, up to one half of these children died. Persistent crying has been reported as a presenting sign (Listernick and Tomita 1991). Some authors have called this group of symptoms in the newborn with myelomeningocele the infantile brainstem syndrome.

Prognosis and complications

Complications are related to severity of injury, quality of assessment and management, and technical expertise of surgical repair. There are reports of symptoms secondary to Chiari malformation stabilizing or improving with time, including newborns with myelomeningocele and symptomatic Chiari malformation. Craniocervical decompression may improve functional outcome (Bindal et al 1995; Pollack et al 1996). Bone regrowth and recurrence of symptoms have also been reported (Hudgins and Boydston 1995; Teo et al 1997). Posterior reversible encephalopathy syndrome has been reported in a patient with Chiari I malformation (Hansberry et al 2013). Other complications that may occur are cough syncope, uveitis, and central sleep apnea (Eken et al 2012; Kitamura et al 2014; Mangubat et al 2014). There have been reports of spontaneous resolution of Chiari malformation and syringomyelia (Briganti et al 2013; Tortora et al 2012).

Arachnoid pathology in Chiari I malformation has an impact on clinical symptoms and postoperative results. Decompressions with arachnoid dissection and an alloplastic duraplasty performed by surgeons experienced with this pathology offer a favorable long-term prognosis (Klekamp 2012). Analysis of a large nationwide health care network showed recently increasing rates of Chiari malformation decompression in children and adults over the past 14 years (Wilkinson et al 2016). Studies in pre- and postoperative diffusion tensor imaging as a tool to evaluate outcome of decompression surgery suggest a normalization of white matter integrity not apparent in conventional imaging (Abeshaus et al 2012). Disability scores such as the Chicago Chiari Outcome Scale and intraoperative MRI are also being investigated as potential methods for measuring treatment efficacy (Bandt et al 2012; Hekman et al 2012; Godil et al 2013). Further studies are needed to determine the usefulness of these tools for measuring outcomes in Chiari malformation.

Clinical vignette

A 28-year-old female patient was followed in a neurology clinic for 14 years. She presented at the age of 14 with partial complex seizures. Headaches were not a prominent symptom. Her neurologic exam did not reveal any focal findings. EEG showed bilateral temporal spikes. CT scan of the brain showed moderate ventriculomegaly. MRI revealed a Chiari 1 malformation with tonsillar herniation down to C3. There was also a rather large cervical syrinx.

The patient was stable for the next 12 years. She then began to have frequent headaches that were moderately disabling. She denied a change in hand coordination, gait, or in the ability to chew and swallow.

Her exam remained without focal neurologic findings. There was no nystagmus or papilledema. Her tongue was normal and there was no atrophy of her hand muscles. Her gait and reflexes were normal.

MRI at this time revealed continued tonsillar herniation down to C3. The cervical syrinx was slightly larger. The patient underwent a suboccipital craniectomy, as well as C1 laminectomy and dural decompression. She did well postoperatively with resolution of her headaches.

Her clinical status has remained good for 2 years. She remains headache-free. There has been no increase in the size of her syrinx or ventricles.

Biological basis

Etiology and pathogenesis

Although the etiology of Chiari malformation is not clearly understood, the literature suggests a condition with heterogeneous causes.

Most evidence points to 2 major causes of Chiari malformation. For most patients, Chiari malformation is a congenital malformation, arising from an embryologic defect in the formation of neural and craniovertebral structures. Stovner performed measurements of posterior fossa size and found patients to have smaller posterior cranial fossae (Stovner et al 1993). Although the small posterior cranial fossa appeared to have little or no clinical significance, the author suggested that it might be the primary developmental anomaly. Nishikawa and colleagues suggest underdeveloped occipital enchondrium and basilar invagination as exacerbating overcrowding in the posterior cranial fossa (Nishikawa et al 1997).

There is evidence that altered CSF dynamics may contribute to the formation of an acquired Chiari malformation. MRI techniques may be used to evaluate cerebrospinal fluid dynamics (Armonda et al 1994). Chiari malformation has also been seen after lumboperitoneal shunting.

Herman and colleagues were the first to report symptomatic Chiari malformation type 1 in siblings (Herman et al 1990).

There is a paucity of studies investigating the role of genetics in Chiari malformations. Genome-wide linkage analysis in subjects with Chiari type 1 identified candidate loci on chromosome 15q21.2-q22.3 and chromosome 9q22.31 (Boyles et al 2006). Finally, an increasing number of specific genetic syndromes with radiographic evidence of Chiari malformations are being published (Greally 2006; Rodrigues et al 2009; Mahore et al 2010).

There is a multifactorial nature to the pathogenesis of symptoms and signs referable to Chiari malformation, including a relationship to hydrocephalus, vascular compromise, direct neuronal distortion, and congenital neural malformation. There is a high likelihood that other associated conditions coexist with Chiari malformation and may be important contributors to the pathogenesis of symptoms. This is particularly true for individuals with spina bifida (Azimullah et al 1991).

Morphometric measures of posterior cranial fossa by CT and MRI have revealed clues concerning the pathogenesis of cerebellar tonsillar herniation. Radiographic evidence shows that a greater degree of cerebellar tonsillar herniation is associated with a shorter clivus length, a wider anteroposterior diameter of foramen magnum, and a wider Boogaard angle (Dufton 2011). The mechanisms suggested include cranial constriction, cranial settling, spinal cord tethering, intracranial hypertension; and intraspinal hypotension (Milhorat et al 2010). Researchers have found differences in cerebrospinal fluid velocities and pressures in type I Chiari malformation patients compared to healthy subjects (Shaffer et al 2011). Chiari 1 decompression can be caused by posterior cranial fossa surgery (Dubey et al 2009). Minor head or neck trauma can precipitate the onset of symptoms in a small number of previously asymptomatic patients with Chiari 1 malformations (Wan et al 2008). A diffusion tensor imaging study revealed widespread microstructural abnormalities in white matter in adolescents with myelomeningocele and Chiari 2 malformation, suggesting ventricular dilation may have additional effects on white matter microstructure (Ou et al 2011).

CSF flow modeling in 3D and 4D have emerged as a technique for evaluating characteristics of flow dynamics when comparing typically developing subjects and those with Chiari malformation (Bunck et al 2012; Linge et al 2013). These techniques show promise for improved diagnostic and therapeutic management.

Epidemiology"

The incidence of Chiari malformation is not known. Increased use of diagnostic imaging has shown Chiari malformations to be much more common than previous estimates of one in 1280 births (Meadows et al 2000). Many children born with the condition do not present with symptoms until adulthood. Chiari malformations occur more often in women than in men with a ratio of about 3:2. Age at presentation varies from 6 to 60 years, with mean age of about 40 years. Presentation is earlier if there is syringomyelia. Chiari 2 malformations may be more prevalent in people of Celtic descent.

Prevention

There are no known primary preventive strategies to decrease the occurrence of Chiari malformation. Improved outcome of secondary disability resulting from Chiari malformation is possible if identification is prompt and specialists with experience in management of Chiari malformation are involved. Some studies show open fetal surgery has promise for improvement of Chiari type 2 and neurologic function in patients with meningomyelocele (Gupta et al 2012).

Differential diagnosis

The clinical manifestations of Chiari malformation exist on a spectrum and involve anatomic severity of the lesion. The differential diagnosis includes brain tumor, hydrocephalus, disorders of the craniovertebral junction, chronic meningitis, multiple sclerosis, cervical myelopathy, and traumatic syringomyelia. Associated syndromes presenting with Chiari malformation include neurofibromatosis, holoprosencephaly, epidermal nevus syndrome, craniofacial defects such as Crouzon disease, craniovertebral defects such as achondroplasia, Klippel-Feil syndrome, and Jarcho-Levin syndrome (deSouza et al 2011). Bosemani and Poretti present a review of cases with neuroimaging findings that demonstrate a broad differential diagnosis of cerebellar disruptions presenting with neurodevelopmental disability (Bosemani and Poretti 2016).

Diagnostic workup

The cornerstone of the workup of a suspected Chiari malformation is MRI of the brain, brainstem, and cervical spinal cord. Sagittal views of the brainstem are particularly important in determining the degree of cerebellar and brainstem herniation. CT scanning may show anomalies such as a beaked tectum and cerebellar tissue around the brainstem on axial views, but it cannot adequately identify the degree of herniation, and may miss Chiari malformations (especially type 1). The cervical cord should be imaged to look for an accompanying syrinx or hydromyelia. Individuals with myelomeningocele should have their entire neuraxis imaged because of the high likelihood that tethered cord or hydromyelia are also present. They should also be assessed for uncompensated hydrocephalus.

A study of 156 patients found that all subjects presenting with a “tonillar blackout sign” (TBS) had Chiari malformation type 1 (Ucar et al 2014). TBS is highly suggestive of Chiari malformation type 1 and is potentially useful in differentiation of symptomatic and asymptomatic Chiari malformation type 1. Rare cases may present with vocal cord palsy causing stridor (Yousif et al 2016).

Based on MRI findings, it has been found that most patients with spinal symptoms had syringomyelia, and a number of individuals with significant tonsillar herniation were clinically asymptomatic (Elster and Chen 1992). It appears that most patients with severe herniation (below the second or third cervical vertebrae) are symptomatic. Cine-MR flow imaging has provided new insight as to the dynamic process involved in the evolution of this pathophysiology (Martin et al 2013; McVige and Leonardo 2014). Microstructural tissue alterations seen on DTI may be present in Chiari malformation type 1 (Eshetu et al 2014). Additionally, there may be a specific role for the middle cerebellar peduncles in Chiari malformation type 1 (Eshetu et al 2014).

Deformities of the midbrain can be seen with Chiari malformation type 1 and mistaken for tumor on CT scan (Hunter et al 1992); however, this is an unusual finding. Type 1 Chiari malformation appears mainly to be associated with syringomyelia and craniovertebral changes, whereas type 2 Chiari malformation has many associated intracranial findings, including breaking of the midbrain tectum, caudal displacement of the fourth ventricle, large massa intermedia, kinking of the lower brainstem, craniolacunia, and hypoplasia of the falx with interdigitation of the cerebral gyri. The degree of tectal beaking has been shown to be associated with the severity of nystagmus in Chiari type 2 (Tubbs et al 2004a). Ultrasonography can be used to identify the presence of type 2 Chiari malformation prenatally, especially when accompanied by spinal dysraphism by detecting the so-called lemon and banana signs (Van den Hof

et al 1990).

Sometimes neurophysiologic tests, such as brainstem auditory response and somatosensory-evoked potential, are useful adjuncts in evaluating patients who appear to be symptomatic from a Chiari malformation.

Fetal MRI features related to Chiari malformation are being investigated and may add to ultrasound, with more specificity for abnormalities such as the degree of downward displacement of cerebellum, signal changes within brain parenchyma, and the type of meningocele (Righini et al 2011).

A whole genome study of Chiari malformation type 1 showed that clustering analyses resulted in the significant identification of patient classes (subtypes), with the pure biological classes derived from patient blood and dura mater samples demonstrating the strongest evidence. Those patient classes were further characterized by identifying enriched biological pathways, as well as correlated cranial base morphological and clinical traits. These results implicated several strong biological candidates, warranting further investigation from the dura expression analysis, and the results also identified a blood gene expression profile corresponding to a global downregulation in protein synthesis (Markunas et al 2014).

Management

An organized approach is essential to comprehensive management of this group of disorders. Management of Chiari malformation relates to 2 primary issues: (1) surgical management of the primary disorder and (2) management of neurologic impairment and disability. Specialists with experience in management of Chiari malformations may improve outcome. Renewed effort to address current deficits in Chiari research involves work groups targeted at pathophysiology, symptoms and diagnosis, engineering and imaging analysis, treatment, pediatric issues, and related conditions (Labuda et al 2011).

One study reported that the overwhelming majority of Chiari I malformation patients (92.9%) managed conservatively do not experience clinical or radiological progression, and a sizeable minority (41.7%) of those who present with symptoms improve (Pomeraniec et al 2016). However, appropriately selected symptomatic patients (sleep apnea and dysphagia) and those presenting with syringomyelia should be considered surgical candidates because of the high rates of clinical (75%) and radiological improvement (87.5%).

The Chiari Health Index for Pediatrics (CHIP) is a patient-reported, Chiari I malformation–specific health-related quality of life instrument, with construct validity in assessing pain-, cognitive-, and emotion-related quality of life, as well as symptomatic features unique to Chiari I malformation (Ladner et al 2016). It holds promise as a discriminative health-related quality of life index in Chiari I malformation outcomes assessment.

There continues to be controversy regarding the optimal surgical technique to address Chiari malformations. The recommended treatment for Chiari I malformations consists of decompressive suboccipital craniectomy and duraplasty when abnormal cine-flow MRI is observed preoperatively and blockage of CSF flow persists intraoperatively despite bony decompression. In order to avoid unnecessary surgical procedures, proper diagnostic studies and patient selection are needed to optimize outcomes (Baisden 2012; Siasios et al 2012).

With regard to surgical management of the primary disorder, issues to consider are: the need for and degree of decompression of the posterior fossa; management of accompanying syrinx of hydromyelia; concurrent craniovertebral anomalies such as basilar impression or invagination (Tominaga et al 1991); associated problems such as hydrocephalus; and shunting or shunt replacement. Optimal neurosurgical management of Chiari malformation is evolving. Advancements in techniques such as endoscopic suboccipital craniectomy and upper cervical laminectomies have made Chiari decompression in pediatric populations comparable with the conventional procedure in terms of minimal surgical invasiveness, recovery time, and complexity of the procedure (Di 2009). Many patients improve with decompression and release of adhesions in the posterior fossa (Dyste et al 1989; Vaquero et al 1990; Bindal et al 1995). Dyste and colleagues were able to develop a predictive model showing the presence or absence of atrophy, ataxia, and scoliosis at the time of the preoperative examination allows prediction of long-term outcome for type 2 Chiari malformation patients (Dyste et al 1989). Nohria and colleagues showed a positive correlation between improvement in hydromyelia or syringomyelia and improvement in signs and symptoms after decompression. The effect of Chiari surgery on scoliosis was inconclusive in that series (Nohria and Oakes 1990). Cases with an associated syrinx may also need to be managed with shunting of the cyst, either with a syringosubarachnoid or shunt, particularly

if the syrinx is large. Neurosurgical intervention for symptomatic syringomyelia associated with Chiari malformation is generally considered an effective and safe treatment, with a 90% chance of long-term stabilization or improvement (Zhang et al 2008; Aghakhani et al 2009). Repeat decompression surgery is sometimes needed (Sacco and Scott 2003). Fetal surgery for myelomeningocele may improve hydrocephalus and hindbrain herniation associated with Chiari 2 malformation (Hirose and Farmer 2009). There is little evidence fetal repair of myelomeningocele improves neurologic function. Further studies are required in the area of fetal surgical repair, safety, and outcome.

In a large surgical series of pediatric and adult Chiari malformation between 1965 and 2013, the postoperative neurologic outcomes were as follows: 75% improved, 17% showed no change, and 9% experienced worsening (Arnautovic et al 2014). Postoperative headaches improved or resolved in 81% of the patients, with a statistical difference in favor of the pediatric series. Postoperative complications were reported for 41% of the series, most commonly with CSF leak, pseudomeningocele, aseptic meningitis, wound infection, meningitis, and neurologic deficit, with a mean complication rate of 4.5%. Complications were reported for 37% of pediatric, 20% of adult, and 43% of combined series. Mortality was reported for 11% of the series. No difference in mortality rates was seen between the pediatric and adult series (Arnautovic et al 2014). Ventral canal encroachment may explain the symptomatology of select patients with type 1 Chiari malformation. Patients with grade I pB-C lines2, with increased ventral canal obstruction, may experience a higher likelihood of syrinx reduction and headache resolution from decompressive surgery with duraplasty than those with grade 0 pB-C2 lines (Ladner et al 2014).

In a survey of the pediatric section of the American Association of Neurological Surgeons, it was determined that there was substantial agreement that surgery should not be carried out on asymptomatic patients and that surgery was indicated for the treatment of brainstem dysfunction, cranial nerve dysfunction, hydromyelia, and scoliosis associated with these malformations (Haines and Berger 1991). There was substantial disagreement about lesser indications for surgery. This continues to be the case (Tubbs and Oakes 2004). Also, as pointed out in this evidence-based review, the role of shunt malfunction and hydrocephalus in producing “Chiari-like” symptoms continues to be an issue that should be pursued on a case-by-case basis.

Many symptoms of Chiari I patients can be conservatively managed (nonoperative) and improve or remained unchanged over time. In 1 study, an overall 67% of pediatric patients had improved cough headache, and 71% had improvement of migraines/diffuse headaches (Killeen et al 2015). The presence of cough headaches is a significant negative predictor of concomitant symptom improvement. This further validates the view that patients with cough headaches should be considered for surgical intervention and provides useful information to counsel patients (Killeen et al 2015).

There is a role for medical management of the neurologic deficits of Chiari malformation. Clearly, symptoms that persist for months after decompression should be treated. A low threshold should be maintained for repeat imaging to investigate whether postoperative changes are playing a role in new or persistent symptoms. Persistent apnea should be evaluated and treated. Case reports documented therapeutic effectiveness of acetazolamide in hindbrain hernia headache and Chiari 1 malformation (Chalaupka 2000; Vaphiades and Braswell 2007).

Outcomes

Chiari type 1 deformity is commonly seen in pediatric neurology, neuroradiology, and neurosurgery and may have various clinical presentations depending on patient age (Poretti et al 2016). In addition, Chiari type 1 deformity is increasingly found by neuroimaging studies as an incidental finding in asymptomatic children. An accurate and reliable selection of patients based on clinical and neuroimaging findings is paramount for the success of neurosurgical treatment (Poretti et al 2016).

Special considerations

Pregnancy

A case report of a 30-year-old woman with a Chiari malformation, presenting with severe pregnancy-induced hypertension, suggests the need for caution and close monitoring (Semple and McClure 1996). Particular concerns for those with an associated syrinx suggest the need for caution, but successful delivery without complications is possible (Parker et al 2002). Although there is a paucity of literature for obstetric and anesthetic management of women with Chiari malformation, spinal anesthesia has been safely used in most surgically decompressed cases (Landau et al

2003; Bag et al 2012). A team approach may be beneficial in complicated cases of surgical management such as cesarean birth pregnancies (Ghaly et al 2012).

Anesthesia

Proper management includes avoidance of increased intracranial pressure and respiratory dysfunction (Nel et al 1998; Radhakrishna 2000). Patients can also have autonomic dysfunction (Ireland et al 1996).

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**References especially recommended by the author or editor for general reading.

Former authors

Stephen L Kinsman MD (original author)

ICD and OMIM codes

ICD codes

ICD-9:
Chiari malformation Type 1: 348.4
Chiari malformation Type 2: 741.00-03
Chiari malformation Type 3: 742.0
Chiari malformation Type 4: 742.2

ICD-10:
Arnold-Chiari syndrome: Q07.0

OMIM numbers

Chiari malformation type I: 8420
Chiari malformation type II: %207950

Profile

Age range of presentation

0-01 month
01-23 months
02-05 years
06-12 years
13-18 years
19-44 years
45-64 years
65+ years

Sex preponderance

male=female

Family history

family history may be obtained

Heredity

heredity may be a factor

Population groups selectively affected

none selectively affected

Occupation groups selectively affected

none selectively affected

Differential diagnosis list

brain tumor
hydrocephalus
disorders of the craniovertebral junction

chronic meningitis
multiple sclerosis
cervical myelopathy
traumatic syringomyelia

Associated disorders

Achondroplasia
Epidermal nevus syndrome
Holoprosencephaly
Hydrocephalus
Infantile brainstem syndrome
Jarcho-Levin syndrome
Klippel-Feil syndrome
Myelomeningocele
Neurofibromatosis
Spina bifida
Spinal dysraphism
Syringomyelia

Other topics to consider

Hydrocephalus
Klippel-Feil syndrome
Myelomeningocele
Nystagmus