Chiari type 1 deformity

Chiari type 1 deformity is a hindbrain disorder associated with elongation of the cerebellar tonsils, which descend below the foramen magnum into the spinal canal.

Defined as cerebellar tonsillar herniation ≥ 5 mm below the foramen magnum.

The hindbrain is not malformed but deformed. Accordingly, “Chiari type 1 deformity,” not “Chiari type 1 malformation” is the correct term to characterize primary tonsillar herniation.

Epidemiology

Chiari type 1 deformity is commonly seen in pediatric neurology, neuroradiology, and neurosurgery and may have various clinical presentations depending on patient age. In addition, Chiari type 1 deformity is increasingly found by neuroimaging studies as an incidental finding in asymptomatic children.

In the past, it was estimated that the condition occurs in about one in every 1,000 births. However, the increased use of diagnostic imaging has shown that CM may be much more common. Complicating this estimation is the fact that some children who are born with the condition may not show symptoms until adolescence or adulthood, if at all. CMs are more prevalent in certain groups, including people of Celtic descent.

A statistically significant (P = .03) female predominance of the malformation was observed, with a female: male ratio of approximately 3:2.

Associated skeletal anomalies were seen in 24% of patients.

Syringomyelia was detected in 40% of patients, most commonly between the C-4 and C-6 levels. Of the 25 patients who presented with spinal symptoms, 23 (92%) proved to have a syrinx at MR imaging. When the syrinx extended into the medulla (n = 3), however, brainstem symptoms predominated. Patients with objective brain stem or cerebellar syndrome had the largest mean tonsillar herniations. Patients with tonsillar herniations greater than 12 mm were invariably symptomatic, but approximately 30% of patients with tonsils herniating 5-10 mm below the foramen magnum were asymptomatic at MR imaging. “Incidental” Chiari I malformations are thus much more common than previously recognized, and careful clinical assessment remains the cornerstone for proper diagnosis and management.
Classification

Chiari type 1 deformity classification.

Etiology

see Chiari type 1 deformity etiology.

Syndromic craniosynostosis

Chiari malformation Type I (CM-I) related to syndromic craniosynostosis in pediatric patients has been well-studied. The surgical management consists of cranial vault remodeling with or without posterior fossa decompression. There were also cases, in whom CM-I was diagnosed prior to the craniosynostosis in early childhood.

A 16-year-old boy who admitted with symptoms related to CM-I. With careful examination and further genetic investigations, a diagnosis of Crouzon syndrome was made, of which the patient and his family was unaware before. The patient underwent surgery for posterior fossa decompression and followed-up for Crouzon's syndrome.

This is the only case report indicating a late adolescent diagnosis of Crouzon syndrome through clinical symptoms of an associated CM-I 4).

Familial clustering

A population-based genealogical resource with linked medical data was used to define the observed familial clustering of Chiari malformation Type I (CM-I). METHODS All patients with CM-I were identified from the 2 largest health care providers in Utah; those patients with linked genealogical data were used to test hypotheses regarding familial clustering. Relative risks (RRs) in first-, second-, and third-degree relatives were estimated using internal cohort-specific CM-I rates; the Genealogical Index of Familiality (GIF) test was used to test for an excess of relationships between all patients with CM-I compared with the expected distribution of relationships for matched control sets randomly selected from the resource. Pedigrees with significantly more patients with CM-I than expected (p < 0.05) based on internal rates were identified. RESULTS A total of 2871 patients with CM-I with at least 3 generations of genealogical data were identified. Significantly increased RRs were observed for first- and third-degree relatives (RR 4.54, p < 0.001, and RR 1.36, p < 0.001, respectively); the RR for second-degree relatives was elevated, but not significantly (RR 1.20, p = 0.13). Significant excess pairwise relatedness was observed among the patients with CM-I (p < 0.001), and borderline significant excess pairwise relatedness was observed when all relationships closer than first cousins were ignored (p = 0.051). Multiple extended high-risk CM-I pedigrees with closely and distantly related members were identified. CONCLUSIONS This population-based description of the familial clustering of 2871 patients with CM-I provided strong evidence for a genetic contribution to a predisposition to CM-I 5).
Pathophysiology

The pathophysiology of CMI is poorly understood and it remains unknown how ICP alterations relate to symptoms and radiological findings.

There is some evidence of impaired intracranial compliance as an important pathophysiological mechanism.\(^6\)

**Magnetic resonance imaging** measurement of transcranial CSF flow and blood flow may lead to a better understanding of the pathophysiology of Chiari malformations and may prove to be an important diagnostic tool for guiding the treatment of patients with Chiari I malformation.\(^7\)

The pathogenesis of a Chiari I malformation of the cerebellar tonsils is grouped into 4 general mechanisms.\(^8\)

It appears that the pathogenesis of Chiari malformation with or without associated basilar invagination and/or syringomyelia is primarily related to atlantoaxial instability. The data suggest that the surgical treatment in these cases should be directed toward atlantoaxial stabilization and segmental arthrodesis. Except in cases in which there is assimilation of the atlas, inclusion of the occipital bone is neither indicated nor provides optimum stability. Foramen magnum decompression is not necessary and may be counter-effective in the long run.\(^9\) It occurs in children and adults. Clinical symptoms mainly develop from alterations in CSF flow at the foramen magnum and the common subsequent development of syringomyelia.

Patients with Chiari malformation type 1 (CMI) often present with elevated pulsatile and static intracranial pressure (ICP).

Several lines of evidence suggest common pathophysiological mechanisms in Chiari malformation Type I (CMI) and idiopathic intracranial hypertension (IIH). It has been hypothesized that tonsillar ectopy, a typical finding in CMI, is the result of elevated intracranial pressure (ICP) combined with a developmentally small posterior cranial fossa (PCF).

The study of Frič and Eide showed comparable and elevated pulsatile intracranial pressure, indicative of impaired intracranial compliance, in both CMI and IIH cohorts, while static ICP was higher in the IIH cohort. The data did not support the hypothesis that reduced PCFV combined with increased ICP causes tonsillar ectopy in CMI. Even though impaired intracranial compliance seems to be a common pathophysiological mechanism behind both conditions, the mechanisms explaining the different clinical and radiological presentations of CMI and IIH remain undefined.\(^10\)

Natural history

see Chiari type 1 deformity Natural history.

Clinical Features
see Chiari type 1 deformity clinical features.

**Diagnosis**

Along with tonsillar herniation, imaging studies have documented additional abnormalities, including smaller and overcrowded posterior cranial fossa. MRI Findings After Surgery for Chiari Malformation Type I is important when evaluating postoperative changes.

Sagittal MRI overestimates the degree of tonsillar ectopia. Misdiagnosis may occur if sagittal imaging alone is used. The cerebellar tonsils are paramedian structures, and this should be kept in mind when interpreting midline sagittal MRI.

**Treatment**

Chiari type 1 deformity treatment.

**Outcome**

Efforts to guide preoperative counseling and improve outcomes research are impeded by reliance on small, single-center studies.

Approximately 1 in 8 pediatric CM-I patients experienced a surgical complication, whereas medical complications were rare. Although complex chronic conditions (CCC) were common in pediatric CM-I patients, only hydrocephalus was independently associated with increased risk of surgical events. These results may inform patient counseling and guide future research efforts.

CM-I in children is not a radiologically static entity but rather is a dynamic one. Radiological changes were seen throughout the 7 years of follow-up. A reduction in tonsillar herniation was substantially more common than an increase. Radiological changes did not correlate with neurological examination...
finding changes, symptom development, or the need for future surgery. Follow-up imaging of asymptomatic children with CM-I did not alter treatment for any patient. It would be reasonable to follow these children with clinical examinations but without regular surveillance MRI.

Outcome assessment for the management of Chiari malformation type 1 is difficult because of the lack of a reliable and specific surgical outcome assessment scale. Such a scale could reliably correlate postoperative outcomes with preoperative symptoms.

Outcome is poor in approximately 3 in 10 patients.

The degree of tonsillar herniation has not been a reliable predictor of either symptom severity or surgical outcome.

Arnautovic et al. identified 145 operative series of patients with CM-I, primarily from the United States and Europe, and divided patient ages into 1 of 3 categories: adult (> 18 years of age; 27% of the cases), pediatric (≤ 18 years of age; 30%), or unknown (43%). Most series (76%) were published in the previous 21 years. The median number of patients in the series was 31. The mean duration of the studies was 10 years, and the mean follow-up time was 43 months. The peak ages of presentation in the pediatric studies were 8 years, followed by 9 years, and in the adult series, 41 years, followed by 46 years. The incidence of syringomyelia was 65%. Most of the studies (99%) reported the use of posterior fossa/foramen magnum decompression. In 92%, the dura was opened, and in 65% of these cases, the arachnoid was opened and dissected; tonsillar resection was performed in 27% of these patients. Postoperatively, syringomyelia improved or resolved in 78% of the patients. Most series (80%) reported postoperative neurological outcomes as follows: 75% improved, 17% showed no change, and 9% experienced worsening. 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 neurological 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.

**Scales**

**Chicago Chiari Outcome Scale (CCOS)**

**Sports**

There is currently no consensus on the safety of sports participation for patients with Chiari I malformation (CM-I).

A prospective survey was administered to 503 CM-I patients at 2 sites over a 46-month period. Data were gathered on imaging characteristics, treatment, sports participation, and any sport-related injuries. Additionally, 81 patients completed at least 1 subsequent survey following their initial entry into the registry and were included in a prospective group, with a mean prospective follow-up period of 11 months.

Of the 503 CM-I patients, 328 participated in sports for a cumulative duration of 4641 seasons; 205 of these patients participated in contact sports. There were no serious or catastrophic neurological
injuries. One patient had temporary extremity paresthesias that resolved within hours, and this was not definitely considered to be related to the CM-I. In the prospective cohort, there were no permanent neurological injuries.

No permanent or catastrophic neurological injuries were observed in CM-I patients participating in athletic activities. The authors believe that the risk of such injuries is low and that, in most cases, sports participation by children with CM-I is safe.23

Systematic review and meta-analysis

Previous studies have attempted to evaluate the utility of preoperative magnetic resonance imaging (MRI) parameters in predicting outcomes in Chiari I malformation. We performed a systematic review and meta-analysis to determine what preoperative imaging features (if any) predict (1) presence of preoperative symptoms or associated findings, (2) need for surgical decompression, or (3) improvement after surgical decompression. METHODS:

All publications through June 2018 on PubMed, Embase, and Cochrane Library databases were searched using the keywords “Chiari I malformation” AND “decompression” OR “imaging.” One thousand two hundred ten publications were identified, and 20 were included for our systematic review; nine were included in the meta-analysis. RESULTS:

Tonsil position, clivus gradient, and scoliotic curve of > 20° were all associated with the presence of preoperative syrinx. Degree of scoliotic curve was associated with length of syrinx. Pre-operative findings of central syrinx morphology, shorter syrinx, and scoliotic curve < 20° were associated with post-operative stability/improvement. Post-operative symptomatic improvement was associated with preoperative pB-C2 line ≥ 3 mm, absence of scoliosis, and presence of syrinx. By meta-analysis, there was no significant difference in post-operative improvement between patients with and without syrinx (OR = 0.89; 95% CI 0.58-1.37). Meta-analysis showed no significant difference in post-operative improvement between patients with and without basilar invagination (OR = 1.31; 95% CI 0.72-2.36).

Multiple studies have attempted to identify preoperative imaging parameters to predict post-operative improvement, but no consistently reliable criteria have been defined. This review and meta-analysis highlight the importance of considering each patient's clinical history and physical exam within the context of associated radiographic abnormalities.24

Case series

see Chiari type 1 deformity case series.

Case reports

see Chiari type 1 deformity case reports.
References


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