Cystic degeneration of the cerebellar tonsils in pediatric patients with Chiari Type I malformation

Clinical article

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Objective. The operative indications and treatment algorithms for pediatric patients with Chiari Type I malformation (CM-I) vary widely. When an intradural approach and duraplasty are thought necessary at the time of surgery, neurosurgeons may elect to fulgurate or resect a portion of the cerebellar tonsils. Histological analyses of cerebellar tonsils resected during decompression in pediatric patients with CM-I revealed multiple abnormal findings including extensive ischemic and degenerative changes. The authors describe an interesting phenomenon of cystic degeneration in the distal ends of the cerebellar tonsils in children undergoing operative treatment of CM-I.

Methods. The authors reviewed the clinical database of 440 pediatric patients who underwent surgical decompression for CM-I performed by a single surgeon. The clinical course, preoperative MR imaging and intraoperative ultrasound characteristics, and histological findings in 3 children found to have tonsillar cystic degeneration were analyzed and detailed.

Results. Cystic changes were subtle but uniformly evident on preoperative MR imaging and were more readily apparent on intraoperative ultrasonography. In each patient, the tonsillar cyst was resected. Histological examination revealed areas of cystic degenerative change characterized by distortion of the normal cerebellar architecture with absent Purkinje and internal granular cell layers. All children experienced improvement in their symptoms, without complication, postoperatively.

Conclusions. Cystic degeneration of the tonsils in pediatric patients with CM-I is an uncommon pathological process most likely resulting from long-standing and excessive compression. Based on their experience, the authors advocate expeditious surgical treatment, including intradural exploration and capacious duraplasty, for patients in whom there is evidence of this phenomenon on preoperative imaging. (DOI: 10.3171/2009.7.PEDS09174)

Key Words • Chiari I malformation • cerebellar tonsils • cyst • ischemia

Chiari Type I malformation is a disorder defined by caudal displacement of the cerebellar tonsils through the foramen magnum and into the upper cervical spinal canal posterior to the dorsal surface of the spinal cord. As a result of this caudal descent, the tonsils typically assume a peglike shape, presumably due to chronic impaction at the CCJ. The pathogenesis and natural history of CM-I remain to be fully elucidated, and as such operative indications and techniques for posterior fossa decompression in these patients vary widely among neurosurgeons. Many surgeons elect to fulgurate or resect a portion of the cerebellar tonsils during decompressive surgery for CM-I. Histological sections of these resected tonsils have demonstrated varied abnormal findings including Purkinje cell loss, atrophic cerebellar cortex, and gliosis with anoxic neuronal changes.3,10 Although the exact cause of these findings is unknown, local trauma and chronic focal ischemia have been implicated in their development.10

We describe cystic degeneration of the distal cerebellar tonsils, a previously unreported finding, in 3 pediatric patients undergoing operative treatment for CM-I. In each case, a discrete cystic-appearing lesion, with MR imaging signal characteristics similar to CSF, was present at
the distal tip of one or both tonsils. We detail the clinical history, pre- and intraoperative imaging characteristics, and histological findings of this rare process and propose potential causative mechanisms.

Methods

We retrospectively identified all pediatric patients who had undergone posterior fossa decompression for CM-I malformation performed by a single surgeon (K.R.C.) at our academic institution between January 1, 1998, and December 31, 2008. The operative reports of these 440 patients were reviewed, and 3 cases were confirmed to involve cystic degeneration of the cerebellar tonsils at surgery. We collected relevant clinical data by reviewing office and inpatient records, pre- and postoperative MR imaging studies, and operative and pathological reports. Approval for this study was obtained from the institutional review board.

Magnetic Resonance Imaging

The patients underwent preoperative MR imaging of the brain and CCJ in a 1.5-T imaging unit. In 2 of the patients, the studies included a sagittal phase contrast sequence performed using a single slice at the midline. Sixteen images were obtained during the cardiac cycle, with phase and magnitude images reviewed in a cine loop. These 2 patients also underwent preoperative dedicated cervical spine MR imaging. In the patient who did not undergo cervical MR imaging preoperatively, the sagittal sequences in the preoperative brain MR imaging study allowed diagnostic visualization of the cervical spinal cord down to the C7–T1 level.

Intraoperative Ultrasound

In each case, intraoperative ultrasound was performed by a neuroradiologist who used a sterilely draped, high-frequency probe. Sagittal and axial images, as well as real-time loops, were obtained in each patient prior to dural incision.

Image Assessment

All imaging findings were retrospectively reviewed by a board-certified neuroradiologist blinded to the site of cystic degeneration identified intraoperatively. These findings were tabulated, with particular emphasis given to the size of the area of cystic change at the distal aspect of the cerebellar tonsil(s), CSF space effacement, tonsillar ectopia below the level of the foramen magnum, cord signal changes, cord morphology, syrinx presence and location, and cine phase contrast sequence findings (CSF pulsatility, tonsillar “pistoning,” and cystic change identification).

Surgical Technique

Posterior fossa decompression was undertaken by performing suboccipital craniectomy as well as laminectomies at C-1 (and C-2 as needed). Following intraoperative ultrasound, the dura mater was opened in typical “Y” fashion, allowing the tonsils to be visualized directly. Cystic lesions were resected in toto and sent to surgical pathology. In each case, duraplasty was performed using a synthetic dural patch and the wound was closed in standard fashion.

Results

Group Description

The 3 patients reviewed in this study were all female, ranging in age from 5 to 7 years. Two of the children initially presented with progressive central sleep apnea, requiring use of CPAP. In addition to apnea, one of the children also had symptoms of caudal brainstem dysfunction including swallowing difficulty with coughing, choking, and spitting. The remaining child suffered from chronic headaches, and MR imaging demonstrated a cervical syrinx. All patients were newly diagnosed and had not undergone previous surgical management of CM-I.

Imaging Findings

All three children underwent noncontrast CCJ and brain MR imaging as described in the Methods section. Descent of the cerebellar tonsils ranged from 2.1 to 2.7 cm (mean 2.4 cm) below the level of the foramen magnum. There were varying degrees of effacement of the dorsal and ventral tonsillar CSF spaces at the CCJ in each patient. Cine phase contrast sequences were acquired in 2 patients and revealed cephalocaudad pistoning of the cerebellar tonsils with effacement of normal CSF pulsatility-related signal changes both ventral and dorsal to the lower brainstem and spinal cord at the foramen magnum. All three patients exhibited kinking of the upper cervical spinal cord at the C-2 level. A definite syrinx, extending from the C5–7 level and having a maximum dimension of 3.2 mm, was identified in one patient (Fig. 1B). Ventriculomegaly was not demonstrated on any of the imaging studies. No vertebral anomalies, skull base osseous anomalies, or other brain parenchymal findings were present.

In each case there was an elliptical region of increased T2 and diminished T1 signal along the margin of 1 or both of the cerebellar tonsillar tips. In 1 of the 3 cases these were noted bilaterally, and in 2 they were found on the right side, corresponding to the location of the most marked tonsillar ectopia. These regions were subtle, especially on T2-weighted images, as they blended with the subjacent CSF spaces. In fact, evaluating the original radiological reports in each case, in only 1 of the cases was the region of cystic change identified by the neuroradiologist interpreting the examination prospectively. They could be identified in all 3 cases retrospectively, however, as CSF signal–like projections from the tonsillar tips (Figs. 1 and 2; see Figs. 6 and 8). In 1 case, deformity of the dorsal dura and posterior spinal canal was noted adjacent to the tonsillar tip cyst, suggesting chronicity (Fig. 1A).

Intraoperative ultrasound demonstrated the cystic areas in each case and was much more definitive than MR imaging. Well-defined, nearly anechoic areas, consistent with cysts, were identified along the tonsillar tips in each patient (Fig. 3; see Figs. 6D and 8D). The intraparenchymal location of the cysts was most apparent on real-time
sonography, with a thin rim of cerebellar tissue seen extending along the cyst surface in each case, and simultaneous motion of the cyst and cerebellum noted on real-time assessment. In each case some degree of effacement of the dorsal and ventral CSF spaces was evident, as was prominent craniocaudal tonsillar pistoning. No Doppler imaging was performed.

Operative Findings and Pathological Examination

In each case, the areas of cystic degeneration at the tonsillar tips were easily identified on durotomy (Fig. 4; see Fig. 7). In contrast to the normal cortical vascular patterns seen along the unaffected areas of cerebellum and tonsils, the areas of cystic degeneration were relatively avascular with a paucity of surface vessels. On excision, gross examination demonstrated a tan-pink, smooth, translucent, uniloculated cyst. Morphological examination demonstrated cerebellar tissue with foci of architectural distortion characterized by cystic degenerative changes (Fig. 5A). Higher magnification of these areas revealed an absence of Purkinje and internal granular cell layers, foci of gliosis (Fig. 5B), and occasional Rosenthal fibers (Fig. 5B inset). Neoplastic changes were absent in all cases.

Postoperative Course

Follow-up data were obtained for the 3 children, with a mean follow-up duration of ~11 months. There were no postoperative complications, including CSF leak or aseptic (chemical) meningitis. The 2 children with lower cranial nerve/brainstem dysfunction quickly experienced significant improvement in symptoms postoperatively, with marked reduction in coughing and choking, no evidence of aspiration on postoperative swallow evaluation, and improvement of their central sleep apnea documented by formal sleep study. The remaining child had interval resolution of her cervical syrinx on routine postoperative imaging.

Illustrative Cases

Case 1

History and Examination. This 5-year-old girl began

![Fig. 1. Case 1. Sagittal T2-weighted MR images of the cervical spine obtained to the right of midline (A) and at the midline (B). A: The right cerebellar tonsil has a cystic appearance as is noted in A (arrowhead), closely following CSF signal intensity. The dorsal dura is bowed along the dorsal aspect of the cystic region (arrows). B: On the midline sagittal image, a dorsal bulge and kink are noted at the cervicomedullary junction (arrow). Regions of high T2 signal are identified in the cord at the C5–7 level, consistent with a syrinx (arrowheads).](#)

![Fig. 2. Case 1. Axial cervical T1-weighted (A and B) and T2-weighted (C and D) MR images at the C1–2 level. Heterogeneous signal (diminished on T1-weighted images and increased on T2-weighted images) is noted in the more cranial aspect of the herniated right cerebellar tonsil (arrows in A and C), consistent with the superior margin of the cystic region. More peripherally, there is isointense cord signal consistent with the noncystic components of the cerebellar tonsil, suggesting that the cyst is intraparenchymal in origin (arrowheads in A and C). More caudally (B and D), the cystic region (arrowheads) is very bright on T2-weighted imaging (D) and hypointense, but not completely CSF-like, on the T1-weighted images (B). The borders are difficult to distinguish from adjacent CSF.](#)

![Fig. 3. Case 1. Sagittal (A) and axial (B) intraoperative ultrasound images obtained after suboccipital craniotomy and C-1 laminectomy, but prior to dural incision. On the sagittal image, marked herniation of the right cerebellar tonsil (T) is noted, with complete effacement of the dorsal CSF space. The prominent cervicomedullary kink (short arrow in A) is again noted, similar to that on the preoperative MR image. A nearly anechoic region is noted along the right cerebellar tonsillar tip (long arrow in A) consistent with a cyst. Although best appreciated on real-time sonography, tissue contiguous with the cerebellar tonsil can be noted along the dorsal aspect of the cyst (arrowhead in A). On the axial image, the spherical anechoic region corresponding to the cyst is again seen (arrows in B). C = cervical spinal cord; OC = operative cavity; T = right cerebellar tonsil.](#)
experiencing frequent occipital and frontal headaches shortly after her 5th birthday. The headaches occurred several times weekly and were most often triggered by exertion, such as playing outside. However, the headaches would occasionally awaken her from sleep at night or early morning. The headaches had become progressively more debilitating, such that she was avoiding outside activities and refraining from play in an effort to prevent them. Her parents denied a history of snoring, and she had no history of gagging, choking, or dysphagia. Magnetic resonance imaging demonstrated CM-I, with the cerebellar tonsils extending ~ 13 mm below the level of the foramen magnum, as well as an associated small syrinx from C5–7 and a cystic region at the distal portion of the right tonsil noted preoperatively (Figs. 1 and 2). The patient’s neurological examination yielded normal findings.

Operation and Postoperative Course. The patient underwent posterior fossa decompression and a C-1 laminectomy. Intraoperative ultrasound confirmed the presence of the cystic structure at the distal end of the right tonsil identified on preoperative MR imaging (Fig. 3). On opening the dura, the cyst was easily recognized (Fig. 4). After resecting the cyst, we coagulated the tonsils and performed dural patch grafting. Postoperatively the child experienced a substantial reduction in the number and severity of her headaches, reporting only occasional mild headaches. Follow-up MR imaging performed at 3 months and again at 9 months postsurgery demonstrated complete resolution of the cervical syrinx (not shown).

Case 2

History. This 7-year-old girl had an ~ 1-year history of worsening sleep apnea ultimately requiring treatment with supplemental oxygen. Formal pulmonary evaluation had included a sleep study that demonstrated > 1000 episodes of central apnea. In addition, her parents reported a 2 to 3-year history of asthma as well as poor feeding, with frequent coughing, choking, and emesis. However, it was not until after demonstration of sleep apnea that an MR imaging study was conducted as part of her evaluation. The imaging study revealed CM-I with the tonsils extending ~ 22 mm below the level of the foramen magnum and significant crowding at the CCJ (Fig. 6A–C).

Examination. The patient had evidence of lower cranial nerve dysfunction, with dysphagia, hoarseness, palatal weakness, and a diminished gag reflex. Other than mild hyperreflexia in her lower extremities (~ 3+ bilaterally), she had no significant long tract findings.

Operation and Postoperative Course. The patient underwent posterior fossa decompression and C-1 and C-2 laminectomies. Following bony decompression, intraoperative ultrasound demonstrated continued crowding at the CCJ and pistoning of the cerebellar tonsils, in addition to a markedly enlarged tonsil on the right with a cystic structure at its distal tip (Fig. 6D). Upon opening the dura, the right tonsil was found to be larger than the left, descending further down into the cervical canal and grossly effacing the spinal cord anteriorly. At its tip was the discrete cystic structure that had been identified on ultrasound but not initially recognized on the preoperative MR study (Fig. 7). The cyst was resected, both tonsils coagulated, and a dural patch sewn into place.

Postoperatively, the child experienced dramatic improvement in her symptoms and, by the time of discharge from the hospital, far less coughing and choking on oral intake. At routine outpatient follow-up ~ 2 weeks after surgery, her parents reported almost complete disappearance of her feeding problems. In addition, a follow-up sleep study performed at 6 weeks postsurgery demonstrated a significant reduction in the frequency and severity of apneic episodes, with no further need for supplemental oxygen.

Case 3

History and Examination. This 6-year-old girl had begun snoring at ~ 2 years of age. Eventually, her parents noted that she had long pauses between breaths while sleeping, and a formal sleep study revealed central apnea. Her symptoms improved after she was started on CPAP at night; however, the level required to adequately control her apneic episodes gradually increased, prompting further evaluation. Magnetic resonance imaging demonstrated CM-I with the tonsils extending ~ 27 mm below the level of the foramen magnum to the level of the C-3 body (Fig. 8). We observed diminished palatal excursion, but the child was otherwise neurologically normal.
Tonsillar cystic degeneration in CM-I

**Operation and Postoperative Course.** Following suboccipital decompression and C-1 and C-2 laminectomies, ultrasound demonstrated marked pistoning of the tonsils as well as cystic structures at the distal tips of the tonsils bilaterally (Fig. 8D). On reinspection, these could be appreciated on the preoperative MR imaging (Fig. 8A–C). After durotomy, the small areas of cystic change were confirmed on the distal ends of both tonsils (not pictured). These were resected, the tonsils coagulated, and a dural patch sewn into place. Postoperatively, the patient had gradual resolution of her symptoms and was no longer requiring CPAP by 3 months postsurgery.

**Discussion**

While theories detailing the pathogenesis of idiopathic CM-I vary widely, the notion that defects of the paraaxial mesoderm result in a congenitally small posterior fossa has gained widespread acceptance, based largely on clinical morphometric studies and laboratory evidence demonstrating undergrown occipital bones in animal models of mesodermal maldevelopment. In a more general sense, CM-I can be said to represent a mismatch between the size of the posterior fossa and its contents, rather than reflecting any particular primary malformation of the involved hindbrain and CNS. Despite this, the neural tissue displaced inferiorly in CM-I, through the bony confines of the foramen magnum and into the upper cervical spinal canal, is subject to abnormal conditions, and histological analyses of cerebellar tonsils resected during decompressive surgery for CM-I have demonstrated multiple abnormal findings.

Koga et al. were the first to report Purkinje cell loss and reactive gliosis in the resected cerebellar tonsils of patients with CM-I. Their series consisted of 4 adult patients, all of whom had syringomyelia, and described similar histological findings in each of the cases. Pueyrredon and colleagues subsequently confirmed these initial findings in a series of 43 pediatric patients with CM-I in whom the tonsils were resected at the time of surgery. They reported histological alterations in 38 of 43 samples, with the remaining 5 specimens showing no evidence of abnormality. The most frequent finding in the series was Purkinje cell loss (present in 32 specimens), followed by gliosis. Additional notable findings included internal granular cell layer loss, focal degenerative changes, and anoxic neuronal changes. Interestingly, Pueyrredon and colleagues also observed that the tonsils in 2 patients with...
acquired CM-I due to chronic lumboperitoneal shunting exhibited the same alterations—namely, Purkinje cell loss and gliosis—as those obtained in patients with congenital CM-I. Taken together, these findings led the authors to conclude that CM-I represents overcrowding of a normally developed hindbrain rather than any primary CNS anomaly. Furthermore, they postulated that the observed histological changes most likely represented a focal phenomenon secondary to specific conditions, namely local ischemia and trauma, resulting from neural tissue abnormally constrained within the narrow confines of the CCJ.10

Purkinje cells have been shown to be especially vulnerable to ischemic insult.6,11 Experimental data suggest that 2 specific properties of Purkinje cells—their reduced capacity to sequester glutamate and an inability to generate energy during periods of relative anoxia—render them particularly susceptible to ischemic death.11 This association between ischemia and Purkinje cell loss may explain the latter’s prominence in the cerebellar tonsils of patients with CM-I. Trapped within the foramen magnum and upper cervical canal, the distal tonsils could potentially undergo chronic ischemic changes resulting from focal constriction of arterial afferents and/or compromise of venous drainage.

Local mechanical trauma to the tonsils as they are subjected to the constant cephalocaudal pistoning of CSF pulsations through the CCJ likely also plays a role in the formation of chronic degenerative changes. Numerous studies have documented findings of cerebellar gliosis and Purkinje cell loss in animal models of blunt head trauma.1,3,7 Alteration in the local microenvironment resulting from compromise of the blood-brain barrier, excitotoxicity, and elaboration of neurotoxic molecules from local reactive microglia have all been speculated to contribute to the observed histological changes. In addition, analysis of rat cerebellar cortex following traumatic injury has demonstrated robust induction of heat shock proteins coinciding with findings of programmed cell death and Purkinje cell loss.1

Formation of a cyst at the distal tip of a cerebellar tonsil lying constrained within the cervical spinal canal, as demonstrated in the 3 cases reported here, represents the culmination of extensive degenerative changes including gliosis and neuronal loss. These histopathological processes are likely the result of focal, chronic ischemic changes as well as repeated local mechanical trauma. The extent of herniation of the tonsils (for example, the level of C-1 compared with C-2 or C-3) in our patient series did not seem to predict or correlate with cyst formation. However, in 2 of the 3 patients found to have cystic degeneration, the cyst developed on the more elongated and inferior lying of the 2 tonsils. In each case, we elected to open the dura and resect the cyst, as well as to fulgurate the inferior portion of the tonsils and perform a capacious duraplasty. We have previously described our technique of utilizing ultrasound to determine the need for dura-
plasty following bony decompression in pediatric patients with CM-I, a technique that we still use. However, the decision to open the dura in these 3 cases was made because, in addition to information obtained from intraoperative ultrasound, the underlying factors thought to be responsible for cyst formation were present. Recognizing that cystic degeneration results from a local environment of long-standing and significant compression, we believe that duraplasty, rather than bone removal alone, is probably necessary to achieve adequate decompression.

Conclusions

This series of 3 patients represents, to the best of our knowledge, the first description of tonsillar cystic degeneration associated with pediatric CM-I reported in the literature. In our experience, this is a relatively rare process resulting from excessive and long-standing compression of the tonsils, leading to focal ischemic changes and characteristic patterns of traumatic injury. The imaging features of cystic degeneration may be subtle but, with careful examination, can be reliably recognized on preoperative MR imaging studies and intraoperative ultrasound. We believe that the presence of tonsillar cystic degeneration on preoperative imaging, similar to the presence of a sizeable or enlarging syrinx, indicates sufficient compression to warrant prompt surgical intervention. By similar reasoning, we propose that an intradural approach with expansive duraplasty is likely needed to achieve adequate decompression. Although the follow-up period is relatively short, data from our small series suggest that these patients experience similarly satisfactory clinical outcomes following decompression as CM-I patients without tonsillar cystic degeneration.

Disclaimer

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

References


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