Asymptomatic Chiari Type I malformations identified on magnetic resonance imaging

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Object. Chiari Type I malformation (CMI) is a congenital disorder recognized by caudal displacement of the cerebellar tonsils through the foramen magnum and into the cervical canal. Frequently, associated findings include abnormalities of nearby bony and neural elements as well as syringomyelia. Cerebellar tonsillar ectopia is generally considered pathological when greater than 5 mm below the foramen magnum. However, asymptomatic tonsillar ectopia is an increasingly recognized phenomenon, the significance of which is poorly understood.

Methods. The authors retrospectively reviewed the records of all brain magnetic resonance (MR) images obtained at our hospital over a 43-month period in an attempt to ascertain the relative prevalence and MR imaging characteristics of asymptomatic CMIs. Of 22,591 patients who underwent MR imaging of the head and cervical spine, 175 were found to have CMIs with tonsillar herniation extending more than 5 mm below the foramen magnum. Of these, 25 (14%) were found to be clinically asymptomatic. The average extent of ectopia in this population was 11.4 ± 4.86 mm, and was significantly associated with a smaller cisterna magna. Syringomyelia and osseous anomalies were found in only one asymptomatic patient.

Conclusions. The authors suggest that the isolated finding of tonsillar herniation is of limited prognostic utility and must be considered in the context of all available clinical and radiographic data. Strategies for treating patients with asymptomatic CMIs are discussed.

KEY WORDS • Chiari malformation • Arnold–Chiari malformation • syringomyelia • syrinx • posterior fossa

The CMs encompass a spectrum of congenital hindbrain herniation syndromes. First described by the Austrian pathologist Hans Chiari in 1891,11 there are at least two accepted variations, frequently referred to simply as CMI and CMII. Although both types characteristically involve some degree of cerebellar herniation;1,7,13,26,30 the CMII is more severe, involving a caudal displacement of the pons, medulla, and fourth ventricle; a strong association with various forms of myelodysplasia, spinal dysraphism, and hydrocephalus; and, almost invariably, an evident and/or symptomatic presentation at or soon after birth, which frequently occurs as a result of associated central nervous system abnormalities (such as myelomeningocele) rather than as a direct result of the CM. The CMI is recognized by caudal displacement of the cerebellar tonsils through the foramen magnum and into the cervical canal. The extent of tonsillar ectopia varies, ranging from a few millimeters to several centimeters. Associated findings include a small posterior fossa, mild caudal displacement of the medulla and/or the fourth ventricle, angulation of the cervicomedullary junction, hydrocephalus, syringomyelia, and various osseous anomalies. Presentation frequently does not occur until the third decade of life.

Although well characterized, the CMI remains poorly understood. Chiari malformation Type I is a congenital, yet slowly dynamic condition, and attempts to understand this condition have had to rely on static examinations across time, usually obtained in patients already symptomatic of their condition. To this end, researchers have examined the extent of tonsillar herniation;1,7,13,26,30 morphological aspects of the posterior fossa;4,29,38,40,44 and pressure differentials across46,47 and CSF motion about35,42,43 the foramen magnum. Despite the contributions of these studies, a basic understanding of the pathogenesis and progression of this condition remains elusive.

This report is meant to serve as a contribution to the understanding of the progression of the CMI. Symptomatic cases have been well characterized.12,21,27,29,41 However, because the CMI typically presents only in middle age, a significant interval remains during which individuals with a CMI are without symptomatic burden. Twenty-one patients are presented here who were incidentally found to have tonsillar herniation significant enough to meet the current criteria for a CMI, but who were without clinical symptoms. The appearance of this malformation on MR imaging is presented, and factors that lead to progression of this condition are discussed in the context of current pathogenetic theories. Finally, strategies for treating patients with asymptomatic CMIs are discussed.

Clinical Material and Methods

We performed a retrospective analysis of MR images
obtained in asymptomatic patients with CMIs. Patients were identified for this study in the following manner (Fig. 1). Using the imaging report database at our institution, all records for MR images of the head and cervical spine that had been obtained from January 1, 1994 through July 13, 1997 were searched for the key words “Chiari,” “syrinx,” and “syringomyelia.” The search string was kept broad intentionally to minimize the possibility of missing relevant records. Results of this initial search were manually reviewed, and patients were excluded if their records suggested tonsillar herniation that was less than 5 mm, there were pathological findings other than a CMI (such as Budd–Chiari syndrome), or reference was made to clinical signs or symptoms. Full medical records were obtained and reviewed in their entirety for all patients not excluded by the aforementioned criteria. Patients were characterized as clinically asymptomatic if they met the following criteria: 1) the patient had no sign or symptom referable to tonsillar herniation (for example, headache or sensory disturbance); or 2) a patient who did have a sign or symptom that could be referable to tonsillar herniation, for example, headache, must have been examined by a neurologist from this hospital who acknowledged the presence of the CM and determined that the patient’s sign or symptom was not the result of the tonsillar herniation. For all patients who met these criteria, an additional search was performed of all hospital documents including admissions notes, clinic visits file, operative notes, and radiological and imaging studies to ensure no exclusion criteria were missed. Those who remained were considered clinically asymptomatic. Magnetic resonance images obtained in these patients were read by the neuroradiologist who had performed the imaging. In cases in which multiple images existed, only the earliest one was used for this study. The MR images were characterized with respect to the following criteria.

**Cerebellar Tonsils.** The cerebellar tonsils were assessed for the degree of herniation and morphological configuration. Tonsillar herniation was measured in a manner similar to that used by others. Briefly, the basion and opisthion were identified, and a line connecting the two was taken to represent the plane of the foramen magnum. Tonsillar herniation was then quantified by measuring a line perpendicular from this line that extended to the most inferior aspect of the cerebellar tonsils visible on all sections. Measurements were made to the nearest millimeter. Additionally, we assessed the general configuration of the tonsils in this study. Because of the difficulty encountered in accurately characterizing specific configurations, the results of this assessment were simply graded as either normal or pointed.

**Osseous Abnormalities.** Magnetic resonance images were assessed for the presence of osseous abnormalities, including cervical or atlantooccipital fusions, basilar invagination, and platybasia. All additional abnormalities were also documented.

**Foramen Magnum.** The greatest sagittal diameter of the foramen magnum was measured in all patients in this study. Because both coronal and axial images were not available in all cases, the transverse diameter of the foramen magnum was not assessed.

**Cisterna Magna.** The relative size of the cisterna magna was used as a surrogate marker for overcrowding of the posterior fossa. Because quantification of this parameter is difficult, the cisterna was qualitatively graded as normal, small, or obliterated.

**Compression of the Medulla.** Compression of the medulla was categorized as either ventral or dorsal, and the compressive structure was recorded.

**Syringomyelia.** Magnetic resonance images in this study were assessed for the presence of syringomyelia to the extent visible on the available images. In cases in which multiple films existed, the earliest set was used.

**Statistical Analysis**

Patient data were transferred to a Microsoft Excel spreadsheet for tracking, comparison, and statistical examination. Tests for association were made using chi-square analysis.

**Results**

Of the 22,591 patients in whom MR images of the head and cervical spine had been obtained at this tertiary care center between January 1, 1994 and July 13, 1997, 175 (0.77%) were found to have CMIs with tonsillar herniation that extended more than 5 mm below the foramen magnum. The proportion of patients in whom the malformations were newly discovered is unknown. Those patients who were symptomatic presented with signs and symptoms common to CMI. Specifically, headache was
The most common complaint, followed by weakness and altered sensation, most commonly paresthesias and dysesthe-sias. Because the presenting signs and symptoms of this disorder have been well documented elsewhere and recently revisited, a more detailed description of symptomatic patients will not be repeated here. The majority of symptomatic patients discovered in this study had either been treated surgically or treatment was pending. According to medical records, only a relative minority of patients with minor symptoms, such as isolated headache, were being observed closely without surgical intervention. Of the 175 patients in whom MR images documented tonsillar herniation extending more than 5 mm below the foramen magnum, 25 (14%) were obtained in patients who were clinically asymptomatic, constituting 0.11% of all patients who had undergone MR imaging of the head and neck. Only one patient among those who met criteria for inclusion in this study had a symptom possibly referable to their CM—headaches—however, in this case the patient was examined by a neurologist who acknowledged the CM and considered the headaches to be consistent with migraines. Magnetic resonance images were available for further review in 21 of 25 asymptomatic patients with tonsillar herniation greater than 5 mm.

This asymptomatic patient population was relatively heterogeneous. The patients’ ages ranged from 1 to 63 years, with an average age of 30 years. There was a nearly even distribution among the sexes (11 male and 10 female patients). Patient characteristics are presented in Table 1, including sex, age at imaging, and reasons for imaging.

The average degree of tonsillar herniation in this group of asymptomatic patients was 11.4 ± 4.86 mm. The range was 7 to 25 mm below the foramen magnum. There was no relationship between tonsillar position and either patient age or patient sex. However, there was a trend for greater tonsillar ectopia in patients with smaller cisternae magna (Table 2). This trend was statistically significant according to chi-square analysis (p < 0.003).

Deformation of the cerebellar tonsils into a pointed or peglike configuration was observed in five patients (25.8%), and was associated with both greater degrees of tonsillar herniation and compression of the brainstem. Unfortunately, CSF flow about the foramen magnum and herniated tonsils could not be directly addressed in this study.

The average size of the foramen magnum of patients in this study was 3.64 ± 0.46 cm, and was not significantly associated with any parameters investigated.

An osseous abnormality, basilar invagination, was observed in only one patient (4.8%). Interestingly, this was the only patient in our study in whom MR images evidenced syringomyelia.

As a surrogate measure of posterior fossa overcrowding, the cisterna magna in each patient in this study was graded as normal, small, or obliterated. The cisterna magna was of normal size in nine (43%), small in eight (38%), or obliterated in four (19%) patients. As noted earlier, there was a trend for greater degrees of tonsillar ectopia with decreasing size of the cisterna magna. No association was found with either patient age or patient sex.

Compression of the medulla was observed in seven patients (33.3%) in this study. Three patients (14.3%) had dorsal compression, which in all cases was caused by the ectopic cerebellar tonsils; one patient (4.8%) had ventral compression by the apex of the dens; and seven patients (33.3%) had both ventral and dorsal compression. As might be expected by the anatomical relationships, compression was associated with greater degrees of herniation, abnormal tonsillar configuration, and smaller sized cisternae magna.

Syringomyelia was found in only one patient (4.8%) and, therefore, significant relationships to other parameters could not be assessed.

### Discussion

In the 109 years since it was first described, considerable effort has been expended in attempts to understand the pathogenesis, progression, and treatment of the CMI. To this end, much progress has been made. Nevertheless, our understanding remains frustratingly incomplete. This study is unique in that it is the first to specifically examine MR imaging characteristics in asymptomatic patients with CMIs. A full understanding of this population is

### Table 1

<table>
<thead>
<tr>
<th>Case No.</th>
<th>Age (yrs),†</th>
<th>Sex</th>
<th>Reason for Imaging</th>
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<tbody>
<tr>
<td>1</td>
<td>42, F</td>
<td>acoustic neuraoma</td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>6, F</td>
<td>precocious puberty</td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>45, M</td>
<td>dizziness / ot posterior BPPV</td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>28, F</td>
<td>diabetes insipidus, hyperprolactinemia</td>
<td></td>
</tr>
<tr>
<td>5</td>
<td>7, M</td>
<td>severe language disorder</td>
<td></td>
</tr>
<tr>
<td>6</td>
<td>31, M</td>
<td>research purposes</td>
<td></td>
</tr>
<tr>
<td>7</td>
<td>25, F</td>
<td>optic neuropathy</td>
<td></td>
</tr>
<tr>
<td>8</td>
<td>3, M</td>
<td>congenital hypothyroidism</td>
<td></td>
</tr>
<tr>
<td>9</td>
<td>1, M</td>
<td>motor delay</td>
<td></td>
</tr>
<tr>
<td>10</td>
<td>45, F</td>
<td>hearing loss</td>
<td></td>
</tr>
<tr>
<td>11</td>
<td>27, M</td>
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<td>12</td>
<td>63, F</td>
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<td>13</td>
<td>27, F</td>
<td>ot acoustic sarcoma</td>
<td></td>
</tr>
<tr>
<td>14</td>
<td>52, M</td>
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</tr>
<tr>
<td>15</td>
<td>54, F</td>
<td>TIA</td>
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<td>44, F</td>
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<td>11, M</td>
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<td>20</td>
<td>33, M</td>
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</tr>
<tr>
<td>21</td>
<td>10, M</td>
<td>research purposes</td>
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* BPPV = benign paroxysmal positional vertigo; TIA = transient ischemic attack.
† Age at imaging.

### Table 2

<table>
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<tr>
<th>Degree of Herniation/No. of Patients</th>
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<tr>
<td>Cisterna Magna</td>
</tr>
<tr>
<td>5–10 mm</td>
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<tr>
<td>10–15 mm</td>
</tr>
<tr>
<td>15–20 mm</td>
</tr>
<tr>
<td>normal</td>
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<tr>
<td>small</td>
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<tr>
<td>obliterated</td>
</tr>
<tr>
<td>9</td>
</tr>
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<td>0</td>
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<tr>
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</tr>
<tr>
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<td>3</td>
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</table>

* Correlation was significant at p < 0.003.
Asymptomatic Chiari Type I malformations

essential if we are to make further progress in understanding how this condition progresses and when to intervene surgically.

**Prevalence of the CMI**

The prevalence of the CMI is unknown and, to our knowledge, has not been formally studied. We found that, within a population undergoing MR imaging at a tertiary care center, 0.77% possessed sufficient tonsillar herniation to meet the standard criteria for the CMI. This proportion, however, must be viewed within the context of the tertiary care setting in which this population underwent imaging. Unfortunately, not all medical records reviewed in this study permitted identification of the date of the initial diagnosis. Therefore, the proportion of these cases that constituted new diagnoses is unknown. Nevertheless, this proportion suggests that the CMI is more common than previously thought. Although this population has not been intentionally examined in any previous study, extrapolation of data from a study by Elster and Chen suggests similar results.

Several, but not all studies have documented a slight female predominance of patients with symptomatic CMI, with female/male ratios ranging from 1.3 to 1.7. We found a nearly equal distribution between the sexes in our population of asymptomatic patients. If there exists a true female predominance among symptomatic patients with the CMI, it is unclear whether this predominance manifests primarily as initial cerebellar ectopia or during subsequent progression of the disorder. It has been suggested that the female cerebellum is “somewhat precocious” in its growth, which might implicate initial ectopia as a cause of this discrepancy between the sexes. At this point, sufficient data to support either supposition are unavailable, and further understanding of this condition and its progression are needed before it is known whether this observation is valid.

**Diagnosis and Herniation**

Current diagnosis of the CMI is confirmed by MR imaging. The generally accepted criterion is demonstrated herniation of one or both cerebellar tonsils extending more than 5 mm below the foramen magnum. The 5-mm cutoff point is the result of several studies of the relative positions of the cerebellar tonsils in healthy volunteers and symptomatic CMI patient populations. However, as MR imaging has become more common, increasing numbers of patients are found with significant degrees of asymptomatic tonsillar herniation. For example, in 1992 Elster and Chen found that, although patients with tonsillar herniations greater than 12 mm were “invariably symptomatic,” approximately 30% of the patients they studied with tonsillar herniations between 5 and 10 mm below the foramen magnum were clinically asymptomatic.

Our findings reinforce and expand on this observation. We found 14% of patients who met the criteria for harboring a CMI to be without clinical symptoms, a proportion that likely represents an underestimate because patients who are symptomatic are more likely to undergo imaging than those who have no symptoms. In contrast to those patients who are already symptomatic from their CMs, asymptomatic patients with this disorder are poorly characterized. Examination of patients within this population may provide important clues to the pathogenesis and progression of this condition and, ultimately, may guide clinical decisions concerning when and how to intervene surgically. To this end, the average extent of tonsillar herniation in this study of asymptomatic patients was 11.4 mm, ranging from 7 to 25 mm. Figure 2 displays a comparison of tonsillar herniation between our patient population and those observed in previous studies of patients with and without symptomatic CMI. Although, by definition, patients with CMs have tonsillar herniation greater than 5 mm, the degree of herniation in our asymptomatic population draws into question the significance of an isolated finding of tonsillar herniation because it is unknown whether these patients are destined to experience progression of the disorder.

One patient in our study was found to have a syringomyelic cavity throughout the length of her spinal cord, which evidenced progression of the disorder, although no symptoms had become manifest. It is our belief that the holocord syrinx found in this patient may have ruptured at its terminal end, thereby relieving any distending pressure.

Why patients with significant tonsillar herniation remain asymptomatic, as well as why those who are symptomatic frequently manifest their symptoms only at adult age, is poorly understood. Numerous theories have been proposed, most of which focus on the role of altered CSF dynamics. Unfortunately, for clinical decision making, noninvasive methods for the detection and consideration of altered CSF flow have been inadequate. The emergence of cine-MR imaging offers new promise as a noninvasive means of assessing CSF dynamics.
The study by Mikulis, et al.,26 of patients who did not have CMs suggested an association between tonsillar ascent with increasing age, but this association is not evident in studies of symptomatic patients with CMIs and was not found in our study population of asymptomatic patients. Although it has been noted that the majority of cerebellar growth occurs after birth,22 the contribution of this growth to normal tonsillar position may be opposed in patients with CMs in whom tonsillar descent is the rule.

Cisterna Magna and Herniation

Relative “overcrowding” of the posterior fossa is well described in patients with CMIs, in whom the posterior fossa is typically small.4,29,38,40,44 Additionally, several18,40 but not all59 morphometric studies have demonstrated a significant correlation between the degree of overcrowding and the extent of tonsillar herniation. The retrospective nature of our study precluded the complex volumetric studies described in previous morphometric studies. We therefore used the relative size of the cisterna magna as a surrogate measure for posterior fossa overcrowding. Assessed in this manner, the size of the cisterna magna was normal in nine patients (43%), small in eight (38%), and obliterated in four patients (19%). The degree of posterior fossa overcrowding was significantly correlated with the extent of tonsillar herniation (Table 2). Despite the consistent finding of posterior fossa overcrowding in patients with CMIs, the pathophysiological significance of this finding is unclear. Several authors have suggested that intrauterine and/or postnatal cerebellar growth within a developmentally small posterior fossa initiates cerebellar tonsillar herniation.4,22,25,38,40,44 A study by Badie, et al.,4 suggested that patients with smaller posterior fossae develop symptoms earlier than those with normal posterior fossae. Thus, although the present evidence remains suggestive of a direct association, future studies are needed to elucidate the relationship between posterior fossa development, tonsillar herniation, and the pathogenesis of this condition. In the meantime, assessment of posterior fossa dimensions in patients with asymptomatic CMs may provide useful data for predicting risk for progression of the disorder.

Tonsillar Herniation and Syringomyelia

The relationship between syringomyelia and the CMI is well documented. Depending on the population studied, 37 to 75% of patients with CMI develop syringomyelia.10,14,25,30,33 However, despite extensive characterization, the precise nature of this relationship remains poorly understood. Several theories have been expounded to explain the development of syringomyelia with CMs. Among the most long-lived have been those of Williams45–47 and Gardner and colleagues,14–16 who postulated craniospinal pressure dissociation and CSF pulsations, respectively, as the pathogenetic processes. These theories have been extensively cited and reviewed, and continue to be popular. More recently, several theories have emerged proposing that increased CSF pressure acts diffusely on the surface of the spinal cord rather than by narrow projection through the central canal. Ball and Dyan11 proposed that CSF, under pressure from subarachnoid obstruction, would pass into the spinal cord by way of Virchow–Rob-
Asymptomatic Chiari Type I malformations

typically enlarged,9,20 it is usually of normal size in cases of CMI9,10,26 and, therefore, unlikely to play a significant role in symptomatic presentation.44 Recognized exceptions are those instances in which osseous anomalies result in posterior displacement of the dens and/or atlantoaxial instability. In such cases, the result is not a narrow foramen magnum per se, but an effectively narrowed posterior fossa outlet. Under such circumstances, it has been found that when the diameter of the canal has been reduced to less than 19 mm, symptoms usually result.24 The average sagittal diameter of the foramen magnum in patients in this study was 3.64 ± 0.46 cm, which is consistent with foramen magnum diameters within the general population of similar age. No patient demonstrated a foramen magnum sagittal diameter less than 19 mm.

Osseous Abnormalities

Numerous osseous anomalies are associated with the CMI. These anomalies include, but are not limited to, a small posterior fossa, basilar impression,13,21,37,38,44 platybasia,21,30,44 fused cervical vertebrae and/or occipitalization of the atlas,13,21,38,44 cervical spin bifida, and craniovertebral junction motion segment abnormalities. An osseous abnormality, basilar invagination, was observed in only one patient (4.8%) in this population of asymptomatic patients. This was the only patient in our study whose images evidenced syringomyelia. The incidence of osseous abnormalities in patients with CMIs has been found to be 23 to 88%.38,44 Although comparisons across studies are difficult, it is of interest that such a small proportion of asymptomatic patients in this study have bone abnormalities.

Strategies for Management of Asymptomatic Patients With CMIs

To date, there have been no clinical studies to provide guidance on the best way to treat asymptomatic patients with CMIs. A recent survey of pediatric neurosurgeons found that the majority of respondents did not believe that their asymptomatic patients with CMIs would ultimately experience symptoms (unpublished data), yet the average age of symptomatic presentation for this disorder is approximately 30 years. Unfortunately, there are no available data to suggest distinct pediatric and adult subpopulations with CMIs. Therefore, it is reasonable to suspect that some proportion of asymptomatic patients will ultimately become symptomatic from their condition. The challenge comes in determining which patients are at greatest risk of progression and when to intervene surgically. The results presented in this study of asymptomatic patients suggest that an isolated finding of tonsillar herniation is an insufficient basis on which to make these determinations. A similar conclusion has been reached in a recent review of symptomatic patients.27

Although our understanding of the CMI remains limited, the available data suggest that a rational approach is possible in treating asymptomatic patients with this condition. We suggest that consideration be given to the extent of tonsillar herniation, the relative CSF flow about the foramen magnum, and the presence or absence of syringomyelia. When taken together with a thorough clinical and neurological evaluation, these variables should provide valuable additional information for clinical decision making for this unique patient population. Thus, when a patient is incidentally found to have a CMI, a reasonable initial evaluation would include a thorough history, neurological evaluation, and further imaging to assess for the presence of syringomyelia and compromised CSF flow about the foramen magnum. Although individual patient characteristics may differ, the presence of neurological signs referable to the patient’s malformation or the presence of a syrinx argue strongly for surgical intervention. In contrast, patients with isolated CMIs without identifiable signs or symptoms and who have no syrinx or compromised CSF flow may be observed closely by using regular neurological examination and periodic imaging. Of course, clinical decision making is most difficult in patients whose clinical presentation lies between these two extremes. Further studies are necessary to ascertain the relative importance of these factors in determining a patient’s risk for clinical progression.

Conclusions

Asymptomatic CMIs are an increasingly recognized finding on imaging studies, the significance of which is poorly understood. We found that within a population of patients who underwent MR imaging at a tertiary care center, 0.77% possessed sufficient tonsillar herniation to meet generally accepted criteria for the CMI. Fourteen percent of these patients were clinically asymptomatic. The degree of tonsillar herniation and the overall MR imaging appearance recognized in this population is similar to that observed in symptomatic patients with this condition. Therefore, we suggest that isolated tonsillar herniation is of limited prognostic utility, and should be considered in the context of all available clinical and imaging data, including assessment of CSF obstruction and the presence or absence of syringomyelia.

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