



Assessing the Prevalence of Ectopic Cerebellar Tonsils and Accompanying Symptoms in Individuals with Various Headaches

ORIGINAL ARTICLE

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ABSTRACT

Objectives: Chiari malformation exhibits well-defined clinical signs, symptoms, and incidence rates in clinical studies. However, cerebellar tonsil ectopia presents with ambiguous symptoms and undetermined incidence rates in numerous studies. Our objective was to determine the incidence of cerebellar tonsil ectopia in individuals with headaches and identify additional clinical symptoms. This aims to improve diagnosis accuracy for clinicians and neurologists, leading to more effective treatment approaches.

Methods: A cross-sectional study conducted in 2022 included 2305 participants aged 4–78 years presenting with headache. Chiari malformation was diagnosed using magnetic resonance imaging (MRI) scans, with a definition of cerebellar tonsil herniation extending beyond 5mm into the cervical canal.

Results: The prevalence of Chiari malformation was 3.4%, with no significant gender difference ($p = 0.72$). There was a significant correlation between Chiari malformation and headache exacerbation due to the Valsalva maneuver ($p < 0.001$) and the presence of vertigo ($p < 0.001$). No significant association was found between Chiari malformation and accompanying symptoms such as nausea ($p = 0.43$), photophobia ($p = 0.2$), phonophobia ($p = 0.52$), and speech disorders ($p = 0.45$).

Conclusion: These findings suggest a notable prevalence of Chiari malformation among headache patients and its association with specific headache characteristics, such as acute and occipital headaches, exacerbation by the Valsalva maneuver, and the co-occurrence of vertigo. These results underscore the need to consider Chiari malformation in the differential diagnosis of patients presenting with these specific headache features.

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OBJECTIVES

Chiari malformations type I (CMs) are classified as rare disorders, either congenital or acquired, marked by the unusual positioning of cerebellar tonsils beyond 5 mm below the spinal canal. This abnormal displacement can disturb the natural flow of cerebrospinal fluid, thereby causing symptoms such as headaches, syrinx, or hydrocephalus. Despite the rarity of this condition, it is significant to understand its implications due to its less than 1% prevalence rate in the United States [1, 2].

Several congenital pathologies, including basilar invagination, platybasia, craniosynocytosis, posterior fossa hypoplasia, atresia of the foramina of Magendie and Luschka, enlarged cerebellar structures, and clival anomalies have been linked with CMs [3–5]. Additionally, congenital biomechanical deficiencies alongside secondary or iatrogenic factors such as trauma, hydrocephalus, tethered cord, and intraspinal hypotension have also been implicated in the etiology of this condition [6–11]. While the majority of CM cases remain asymptomatic in the initial stages, symptoms typically start manifesting between the ages of 25 to 40, ranging from mild headaches and dysesthesias to severe conditions such as respiratory failure and tetraplegia [3, 4].

Interestingly, research has indicated a potential correlation between the length of herniated cerebellar tonsils and the severity of clinical manifestations [3, 12–14]. Given the diverse extent of nervous tissue displacement, variations in the size of the foramen magnum, and the range of structures compressed, the clinical presentation of CMs is highly protean, posing a complex challenge for neurology practitioners. Increasing awareness about the different clinical presentations could foster more prompt recognition of the condition and timely magnetic resonance imaging (MRI) testing [3].

Headaches, arguably the most common manifestation, can sometimes be the sole indication of the presence of CMs or may be completely absent [15, 16]. Characteristically, these headaches, associated with Chiari, are occipital-suboccipital in nature, often exacerbated by Valsalva maneuvers, positional changes, head dependency, and physical exertion [3, 15].

Management strategies for CMs often revolve around decompressive suboccipital craniectomy, though the rationale behind the surgical intervention is a subject of ongoing debate [17]. Generally, conservative management without surgery is adopted for headaches unless the patient experiences significant disability due to a specific occipital-suboccipital headache accompanied by a concurrent cough headache.

This study aims to investigate the prevalence of Chiari malformations (CMs) and cerebellar ectopia in headache patients. By analyzing and discussing the accompanying

symptoms, we aim to enhance understanding, facilitate timely diagnosis, and optimize management strategies for this complex neurological condition.

MATERIALS AND METHODS

STUDY DESIGN AND PARTICIPANT RECRUITMENT

We conducted a cross-sectional study in 2022 using a systematic random sampling method. The study participants were individuals of all ages who presented with headache symptoms and were referred to the MRI department. Exclusion criteria were applied to individuals with known brain disorders (such as tumors, multiple sclerosis, hydrocephalus), neurological symptoms (such as paresis, paresthesia), a history of head trauma, or prior brain surgery. The diagnosis of Chiari malformation was made based on cerebellar tonsil protrusion greater than 5 mm, and borderline diagnosis was determined by a descent of 3–5 mm into the foramen magnum, as observed in the midsagittal plane on the T1 sequence. Please refer to [Figure 1](#) for a visual representation.

HEADACHE DEFINITION AND CLASSIFICATION

The classification of headaches was determined by two factors: acuteness and severity. The severity of headaches was defined as mild (a headache that somewhat interferes with the ability to function), moderate (a headache that hinders the ability to function but does not necessitate rest), and severe (a debilitating headache requiring rest in bed). Headache acuteness was classified as acute (lasting hours up to a day), subacute (lasting a



Figure 1 Sagittal T1-weighted TSE image showing cerebellar tonsils below the level of the foramen magnum.

day up to a month), and chronic (persisting for months or years).

RESULTS

PARTICIPANT DEMOGRAPHICS AND CHIARI MALFORMATION PREVALENCE

In a 2022 study at Vali-Asr and Imam Reza Birjand hospitals, 2305 individuals with headaches underwent MRI examinations. Among them, 79 patients (3.4%) showed cerebellar tonsil herniation exceeding 3 mm into the cervical canal. The prevalence of Chiari malformation and cerebellar tonsil ectopia based on herniation extent beneath the foramen magnum level is as follows:

- Ectopia <3 mm: 1434 females (96.8%) and 792 males (96.2%).
- Ectopia 3–5 mm: 27 females (1.8%) and 19 males (2.3%).
- Ectopia >5 mm: 21 females (1.4%) and 12 males (1.5%).

Overall, 84.9% (1956 patients) had ectopia <3 mm, and 15.1% (349 patients) had ectopia ≥3 mm. Importantly, gender did not significantly influence the distribution of ectopia lengths ($p = 0.72$).

HEADACHE INTENSITY

A comparison of headache severity across healthy individuals, those with cerebellar tonsillar ectopia, and individuals diagnosed with Chiari malformation is provided in Table 1. The majority of patients from all three

	EXTENT	HEADACHE SEVERITY		
		MILD	MODERATE	SEVERE
Ectopia	<3 mm	172 (7.7%)	1859 (83.5%)	195 (8.8%)
	3–5 mm	4 (8.7%)	36 (78.3%)	6 (13.0%)
	>5 mm	3 (9.1%)	25 (75.8%)	5 (15.2%)
Total		179 (7.8%)	1920 (83.3%)	206 (8.9%)

Table 1 Comparison Of The Relative Frequency Of Cerebellar Tonsil Ectopia According To Severity.

	EXTENT	LOCATION			
		FULL HEAD	OCCIPUT	FRONTAL	OTHER
Ectopia	<3 mm	1072 (48.2%)	274 (12.3%)	690 (31.0%)	190 (8.5%)
	3–5 mm	17 (37.0%)	18 (39.1%)	8 (17.4%)	3 (6.5%)
	>5 mm	2 (6.1%)	29 (87.9%)	1 (3.0%)	1 (3.0%)
Total		1091 (47.3%)	321 (13.9%)	699 (30.3%)	194 (8.4%)

Table 3 Comparison Of The Frequency Of Ectopia Of Cerebellar Tonsils According To Location Of Headache.

cohorts reported experiencing moderate headaches, with no substantial difference noted ($p = 0.57$).

HEADACHE PERIOD

The study assessed the duration of headaches, noting varied results across different groups. Table 2 denotes the frequency of cerebellar tonsil ectopia corresponding to headache occurrence timing. Most Chiari malformation patients reported acute headaches (69.7%), while those with cerebellar tonsillar ectopia and the healthy group primarily reported subacute headaches (39% and 70%, respectively). The distinction between the Chiari malformation group and the cerebellar tonsillar ectopia group was statistically significant ($p < 0.001$).

HEADACHE LOCALIZATION

Headache localization varied significantly among the groups, as displayed in Table 3, which details the cerebellar tonsil ectopia frequency based on headache location. The majority of Chiari malformation patients experienced occipital headaches (87%), while cerebellar

	EXTENT	HEADACHE PERIOD		
		ACUTE	SUBACUTE	CHRONIC
Ectopia	<3 mm	284 (12.8%)	1558 (70.0%)	384 (17.3%)
	3–5 mm	16 (34.8%)	18 (39.1%)	12 (26.1%)
	>5 mm	23 (69.7%)	5 (15.2%)	5 (15.2%)
Total		323 (14.0%)	1581 (68.6%)	401 (17.4%)

Table 2 Comparison Of Ectopic Frequency Of Cerebellar Tonsils According To Time.

	EXTENT	EXACERBATION BY VALSALVA MANEUVER	
		NEGATIVE	POSITIVE
Ectopia	<3 mm	1930 (86.7%)	296 (13.3%)
	3–5 mm	20 (43.5%)	26 (56.5%)
	>5 mm	6 (18.2%)	27 (81.8%)
Total		1956 (84.9%)	349 (15.1%)

Table 4 Comparison Of Frequency Of Ectopia Condition According To Exacerbation By Valsalva Maneuver.

tonsil ectopia patients (39%) and healthy individuals (12%) reported headaches in diverse locations. This variation was statistically significant ($p < 0.001$).

VALSALVA MANEUVER-INDUCED HEADACHE EXACERBATION

Table 4 examines the correlation between Chiari malformation, cerebellar tonsil ectopia and normal population and headache exacerbation triggered by the Valsalva maneuver. A significantly higher percentage of Chiari malformation (81.8%) and cerebellar tonsil ectopia patients (56.5%) reported positive Valsalva maneuver responses compared to healthy participants (13.3%) ($p < 0.001$).

ACCOMPANYING CLINICAL SYMPTOMS WITH HEADACHE

Table 5 examined the occurrence of accompanying symptoms such as nausea, photophobia, phonophobia, and speech disorder in relation to headache, finding no significant differences among the three groups ($p = 0.43$, $p = 0.2$, $p = 0.52$, and $p = 0.45$, respectively). Furthermore, patients with Chiari malformation had a significantly higher incidence of vertigo (75.8%) compared to those with cerebellar tonsillar ectopia (17.4%) and healthy individuals (18%) ($p < 0.001$).

DISCUSSION

Our comprehensive study conducted in 2022 involved a cohort of 2305 patients, with the primary objective of analyzing the prevalence, extent, and accompanying symptoms of Chiari malformation.

Our research, along with two other studies, investigated headache types and related symptoms in various patient groups. We found no significant differences in common symptoms among groups, implying these may not be key indicators for Chiari malformation or cerebellar tonsillar ectopia. However, a strong connection between cerebellar tonsil ectopia frequency and vertigo, particularly in Chiari patients, was discovered. This suggests the importance of considering non-traditional symptoms for diagnosis and treatment.

Additionally, a study indicated a range of symptoms and an inverse relationship between tonsillar ectopia size and typical Chiari symptoms, implying a correlation between condition and symptom severity [18, 19].

This study revealed that patients with <3 mm had higher pain scores than those in the 3–5 mm or >5 mm groups, suggesting varied headache severity in different groups. These results imply that conditions like Chiari malformation and cerebellar tonsillar ectopia can influence symptom severity, but no clear correlation was found with nausea and photophobia. There could, however, be a significant link between these conditions and symptoms like vertigo and certain headaches. More research is needed to improve symptom management understanding.

Our findings align with previous research [20–22] and demonstrate a significant association between Chiari malformation or cerebellar tonsil ectopia and exacerbated headaches during the Valsalva maneuver. Our results indicate a similar trend, with a higher proportion of patients experiencing increased headaches compared to healthy participants. Furthermore, our study reinforces the importance of distinguishing between headaches associated with Chiari malformation and primary cough headaches. These insights enhance our understanding of symptom management and contribute to more accurate differential diagnosis.

Our study revealed a strong association between the frequency of ectopia and the occurrence of vertigo. Chiari malformation patients had a significantly higher incidence of vertigo compared to those with cerebellar tonsillar ectopia and healthy individuals. Another study also found vertigo to be a common symptom in patients with symptomatic tonsillar ectopia, along with unstable gait and limb numbness. These findings indicate that neuroanatomical variations play a crucial role in the manifestation of vertigo. Consequently, there is potential for a targeted approach to diagnose and manage vertigo in patients with Chiari malformation and cerebellar tonsillar ectopia [23].

Our study, along with previous research, sheds light on the correlation between symptoms like nausea, photophobia, phonophobia, speech disorder, and headaches in three distinct groups categorized by the

ECTOPIA		NAUSEA	PHOTOPHOBIA	PHONOPHOBIA	SPEECH DISORDER	VERTIGO
<3 mm	Negative	1514 (68.0%)	1915 (86.0%)	1950 (87.6%)	2104 (94.5%)	1825 (82.0%)
	Positive	712 (32.0%)	311 (14.0%)	276 (12.4%)	122 (5.5%)	401 (18.0%)
3–5 mm	Negative	32 (69.6%)	41 (89.1%)	41 (89.1%)	43 (93.5%)	38 (82.6%)
	Positive	14 (30.4%)	5 (10.9%)	5 (10.9%)	3 (6.5%)	8 (17.4%)
>5 mm	Negative	19 (57.6%)	25 (75.8%)	31 (93.9%)	30 (90.9%)	8 (24.2%)
	Positive	14 (42.4%)	324 (14.1%)	283 (12.3%)	3 (9.1%)	25 (75.8%)

Table 5 Accompanying Clinical Symptoms with Headache.

degree of ectopia. Consistent with earlier studies, we found no significant differences in these symptoms among the groups, indicating a universal occurrence rather than a connection with ectopia severity. However, when examining the association between ectopia frequency and vertigo incidence, our data aligns with previous research. Patients with Chiari malformation, characterized by greater ectopia, had a significantly higher incidence of vertigo compared to those with cerebellar tonsillar ectopia and healthy individuals. This emphasizes the importance of understanding the symptom profile in patients with varying degrees of ectopia, with vertigo being particularly linked to severity. Therefore, it is crucial to focus on effective symptom management, especially for vertigo in severe cases like Chiari malformation [24, 25].

One notable limitation of this study lies in its cross-sectional design, which precludes establishing a cause-effect relationship between the observed symptoms and the presence of Chiari malformation or cerebellar tonsillar ectopia. The findings of this study are also contingent upon the reliability of participant self-reporting during interviews, which can be subject to recall bias or inaccurate reporting. Furthermore, the exclusion criteria applied may have potentially left out certain symptomatic cases with pre-existing brain disorders or prior brain surgeries.

CONCLUSION

Our study found that symptoms were similar among patients with Chiari malformation and cerebellar tonsillar ectopia, except for a higher occurrence of vertigo in Chiari malformation cases. This highlights the need for a nuanced approach to diagnosis and management. Our results align with other studies, emphasizing diverse headache severity and the impact of physical manifestations on symptoms. We also discovered the significance of the Valsalva maneuver in triggering headaches, distinguishing them from benign cough-associated headaches. This correlation suggests a targeted approach to diagnosis and management. Overall, our findings stress the importance of understanding symptoms and effective management strategies for these conditions.

DATA ACCESSIBILITY STATEMENTS

Prior to undergoing MRI scans, an interview was conducted with each participant by a trained radiology department technician. Following the interview, a checklist-based questionnaire was administered, which consisted of two sections: demographic information

and research-specific characteristics. All data obtained from the participants was systematically recorded in a structured format.

STATISTICAL ANALYSIS

The collected data was analyzed using SPSS software version 25 (IBM Corp. Released 2017). Parametric variables were analyzed using Chi-square tests and Fisher's exact test when needed, with a significance level of 0.05.


ETHICS AND CONSENT


The research protocol, including proposal development, data collection, and final reporting, was closely supervised and approved by the local ethics committee affiliated.


COMPETING INTERESTS

The authors have no competing interests to declare.


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