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CHIARI MALFORMATION AS A DIFFERENTIAL DIAGNOSIS OF RECURRENT SYNCOPE

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ABSTRACT

Arnold-Chiari malformations are rare anomalies involving inferior displacement of the cerebellar tonsils and hindbrain through the foramen magnum. We present the case of a 19-year-old woman with a relatively rare presentation of recurrent syncope. The patient underwent surgical treatment with resolution of the symptoms. Few cases of surgical treatment have been described. This case underscores the importance of considering Chiari malformations in the differential diagnose of a common symptom - recurrent syncope - and the relevance of considering these patients for surgical treatment.

KEYWORDS

Arnold-Chiari syndrome, Chiari malformation type I, syncope

LEARNING POINTS

- Chiari malformation type I (CM-I) should be included in the differential diagnosis in recurrent syncope, especially if accompanied by headache, vertigo, or transient sensory or motor symptoms in the arms or legs.
- In patients with CM-I with syncope, the good clinical results of surgical decompression should be kept in mind.

INTRODUCTION

Arnold-Chiari malformations are a group of deformities first described by the pathologists Julius Arnold and Hans Chiari[1]. The condition involves the posterior fossa and hindbrain. It consists of a spectrum of abnormalities that vary from herniation of the cerebellar tonsils through the foramen magnum to a complete absence of the cerebellum. Additional intracranial or extracranial anomalies may also be present[2]. Four distinct types are described according to the type of brain tissue herniation into the spinal canal and the specific developmental abnormalities of the brain or spine[3]. Chiari malformation type I (CM-I) is the most common form but it is also the least severe. It is defined by the herniation of one or both cerebellar tonsils 5 mm or more below the foramen magnum, a measurement taken using the McRae line (from the basion to the opisthion). This causes direct compression of neurological structures and the upper spinal cord, resulting in cerebrospinal fluid (CSF) obstruction and potentially syringomyelia[2]. Often asymptomatic, it typically manifests in adulthood[1,4]. Occipital/cervical pain

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or headache is the most frequent symptom and worsens with the Valsalva manoeuvre due to the reduced size of the foramen magnum[1]. Syncope is a relatively uncommon symptom, and the precise cause of CM-1-related syncope remains poorly understood[4].

CASE DESCRIPTION

A 19-year-old female with no significant medical or surgical history presented to the emergency department after experiencing syncope, without associated head trauma, following physical activity. She reported recurrent episodes of syncope without prodromal symptoms and episodes of dizziness. She denied taking any regular or occasional medication, alcohol consumption, or drug abuse.

Upon admission, the patient was hemodynamically stable (blood pressure of 112/55 mmHg and heart rate of 60 bpm), eupnoeic, with oxygen saturation (SpO_2) of 99% on room air, and afebrile. The physical examination did not reveal significant findings, with normal cardiac and pulmonary auscultation, and no abdominal abnormalities. There were no oedema or signs of inflammation in the lower limbs. Her electrocardiogram and initial laboratory tests, including blood glucose, were within normal limits. Given the history of recurrent syncope, the patient was admitted for further investigation.

During hospitalization, the patient underwent Holter monitoring, echocardiogram, and neck vessel Doppler ultrasound, all of which were unremarkable. Orthostatic hypotension was also ruled out. A detailed neurological examination revealed hypesthesia and loss of pain sensation on the left side of the face and limbs, along with weakness (4/5 strength) in both the left upper and lower limbs on the arm extension test and Mingazzini test. Dysmetria and adiadochokinesia were also noted on the left side. These findings prompted the performance of a cranial computed tomography (CT) scan, which showed downward displacement of the cerebellar tonsils through the foramen magnum, with partial obliteration of the cerebrospinal fluid spaces in a regional area. To clarify these findings, magnetic resonance imaging (MRI) was performed, confirming the presence of cerebellar tonsil ectopia, approximately 6 mm caudal to the McRae anatomical line, leading to the diagnosis of Arnold-Chiari syndrome *(Fig. 1)*. The case was discussed with the neurosurgery team, who proceeded with decompressive surgery. Since the surgery, the patient has not experienced any recurrence of symptoms.

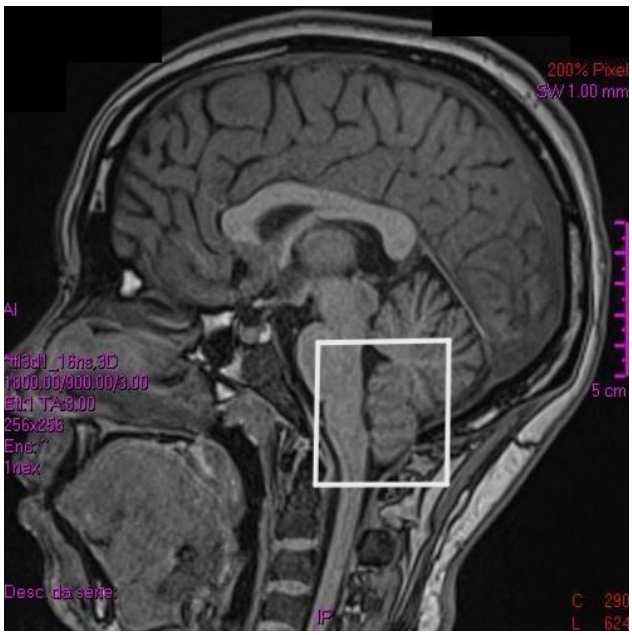


Figure 1. Magnetic resonance imaging of the head shows downward displacement of the cerebellar tonsils through the foramen magnum (white box).

DISCUSSION

Arnold-Chiari malformations are rare anomalies involving inferior displacement of the cerebellar tonsils and hindbrain through the foramen magnum[5,6]. CM-1 is the most common variant and the least severe[1,2]. The caudal descent of the cerebellar tonsils below the foramen magnum results in direct compression of neurological structures within the foramen magnum and upper spinal cord, leading to obstruction of CSF and potentially associated syringomyelia[1,2]. The aetiology is not completely understood. It is likely that more than one mechanism leads to the same Chiari malformation. The displacement of the cerebellar tonsils occurs because of the reduced volume of the posterior fossa, which can be caused by congenital hypoplasia or secondary to acquired morphologic changes like premature closure of sutures, calvarial dysplasia, or genetic/syndromic factors[2]. Classically, patients with CMI present with pain, weakness, sensory loss, dizziness, gait disturbances, and cranial nerve palsies[7]. Syncope is one of the rarest presentations[4,7], that can occur with or without premonitory symptoms[4]. Different mechanisms have been proposed to explain this symptom including transient increase of intracranial pressure during a Valsalva manoeuvre leading to vertebrobasilar artery compression; compression of the midbrain ascending reticular system and compression of cardiorespiratory centres or their efferent/afferent pathways[4]. Some cases of patients with orthostatic syncope have been reported. These cases are also challenging since the tilt test is not always positive, although it is used in diagnosis of neurogenic orthostatic hypotension. However, there is no gold standard test for diagnosis of neurogenic orthostatic hypotension[7]. In the literature, there are also reports of cough, orthopnoea and severe respiratory dysfunction associated with syncope. Central apnoea can be also explained by compression of the ascending reticular system, which is improved by decompressive surgery[7].

Brain imaging is not required for uncomplicated cases of syncope. Moreover, despite of the fact that decompression has led to good clinical outcomes in CM-I with syncope, there are few reported cases of surgical treatment in these patients[7]. Hence, we emphasise the importance of a systematized approach to a common symptom such as syncope, including considering central nervous system

evaluation. Regardless of the low incidence, Chiari malformations should be included in the differential diagnose of recurrent syncope and the good clinical results of surgical decompression should be kept in mind.

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