Chiari I malformation: Case report

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RESUMO
Arnold Chiari type I malformation (CM-I) is a genetic disease first described in 1891 by Hans Chiari (SCHIJMAN, 2004). CM-I is characterized by the descent of the cerebellar tonsils more than 5 mm from the lower margin of the foramen magnum, which can impede the flow of cerebrospinal fluid (CSF) and is often associated with syringomyelia (BALL; CRONE, 1995; ROMERO-LUNA et al., 2022), generating great clinical neurological interest due to the difficulty in diagnosis (MORO et al., 1999).

Palavras-chave: Chiari I malformation, Clinical implications, Case report.

1 INTRODUCTION

Arnold Chiari type I malformation (CM-I) is a genetic disease first described in 1891 by Hans Chiari (SCHIJMAN, 2004). CM-I is characterized by the descent of the cerebellar tonsils more than 5 mm from

the lower margin of the foramen magnum, which can impede the flow of cerebrospinal fluid (CSF) and is often associated with syringomyelia (BALL; CRONE, 1995; ROMERO-LUNA et al., 2022), generating great clinical neurological interest due to the difficulty in diagnosis (MORO et al., 1999).

The clinical picture of CM-I is variable, depending on the age of the patient, with associated anomalies and body structures compromised by the syndrome (ROWLAND; PEDLEY, 2011). It is more common in the female population, a ratio of 3:1 in relation to males (LÓPEZ et al., 2007). Its treatment is complex and is defined according to the malformations presented (BRITO et al., 2019). The most commonly used surgical option is decompression of the posterior cranial fossa (ALZATE et al., 2001; VALE et al., 2014; BRITO et al., 2019).

The objective of the present study is to report a case of Chiari type 1 malformation and to discuss its possible cause and clinical implications.

2 CASE REPORT

A 46-year-old female patient with Chiari type I malformation diagnosed in 2017 underwent posterior fossa decompression surgery in 2021. Preoperative symptoms included: headache, neck pain, upper and lower limb paresis, low back pain, insomnia and paresthesia. Postoperatively, the following complications were recorded: paresthesia in the right upper limb, grade IV paresis in the right upper limb and exhaustible bilateral plantar clonus. As a post-operative improvement, the patient showed improvement in her left upper limb paresis. In order to continue improving the symptoms described, the dose of the drug pregabalin was adjusted to 150 mg every 12 hours and an MRI scan was carried out to assess the cervical spine.

Two months after the posterior fossa decompression surgery, the patient underwent electroneuromyography of all four limbs, which revealed demyelinating involvement of the sensory fibres of the median nerves of the wrist (mild carpal tunnel syndrome bilaterally) and the lower limb examination showed normal parameters.

In the two years following the craniotomy (2022 and 2023), the patient underwent a cranial computed tomography (CT) scan. The CT scan carried out in 2022 showed possible post-therapeutic changes, such as fibrosis in the nape of the neck, and a correlation with the clinical data was requested for confirmation. The 2023 scan showed no pathological calcifications. The examination reports showed no alterations, with signs of occipital craniotomy, brainstem, cerebellum, cerebral parenchyma, ventricular system, sulci and cerebral cisterns with normal dimensions, shapes and aspects.

In 2023, the patient took the following oral medication: Velija 60mg, Topiramate 50mg, Alprazolam 0.5mg, Eszopiclone 3mg, Bupropion Hydrochloride 300mg and Pregabalin 150mg.

In June 2023, the patient underwent a cranioencephalic magnetic resonance imaging scan.

Analysis of the scan showed signs of occipital cranioplasty with no signs of abnormality, no evidence of intraparenchymal hemorrhage, negative diffusion for acute ischemia, no intracranial expansive process, normal aeration of the visible portions of the paranasal air cavities, ventricular system, cerebellum and brainstem with unchanged dimensions, shape and topography.

Despite complementary exams indicating no significant anomalies or alterations, the medical report for the patient, two years after surgery, was that she developed residual clinical symptoms that did not improve after the operation, and suggested retirement due to inability to work.

3 DISCUSSION

Chiari malformations are a group of conditions originally described in 1891 and 1896 by Hans Chiari, a German pathologist. The malformations are classified into 4 types based on the degree of herniation of cerebellar structures and associated lumbar malformations (SCHIJMAN et al., 2004). In CM-I, there is usually descent of the cerebellar tonsils more than 5 mm from the lower margin of the foramen magnum, and it is rarely seen below the second cervical vertebra (C2), and it is not associated with myelomeningocele (PAUL et al., 1983; BALL; CRONE, 1995).

Several theories attempt to explain the origin of this anomaly. Tonsillar ectopia, the main anatomopathological finding of CM-I, can be explained by a disproportion between the space of the posterior cranial fossa and the local nerve tissue (ALDEN et al., 2001; BEJJANI, 2001), resulting from the underdevelopment of the four caudal occipital somites that form the posterior part of the skull base. In this context, the Pax-1 gene stands out, which acts in somite segmentation and sclerotome differentiation (SPEER et al., 2000). In 1979, Marin-Padilla compared the occipital bone to a vertebra, by means of three pairs of somites that function as a rudimentary occipital vertebra (MARIN-PADILLA, 1979). According to the study by Nishikawa et al. (1997), the parts of the occipital endocranium (occipital scales and condyles) are usually underdeveloped, as a cause of occipital somite involvement. In 1991, Marin-Padilla considered CM-I to be a type of dysraphism that differs from other cases in the degree of involvement of the neural crests and, consequently, the Central Nervous System (MARIN-PADILLA, 1991).

The result of these alterations is a small posterior cranial fossa (ALDEN et al., 2001), which predisposes to herniation of its contents. Once cerebellar herniation has occurred, it leads to a dissociation of craniospinal pressure (creation of a gradient), which produces a centrifugal force vector from the cranial cavity towards the intraspinal compartment, which favors the descent of the cerebellar tonsils through the foramen magnum (STOVNER et al., 1993; SAHUQUILLO et al., 1994).

The patient underwent posterior fossa decompression surgery in 2021. Alzate et al. (2001) state that the treatment of CM-I is one of the most controversial topics in neurosurgery, but conclude that bone decompression of the craniovertebral junction is the only strategy that is not under discussion (ABLA et al.,

2010). Batzdorf (1988) suggests that, regardless of the pathophysiology of CM-I, the main objectives of surgical treatment are: (1) to relieve the craniospinal pressure gradient at the craniovertebral junction, (2) to restore the subarachnoid space, (3) to eliminate the syringomyelic cavities, if present, and (4) to decompress the neural structures involved.

In the present report, the patient presented with headache, neck pain, right upper limb paresthesia, grade IV right upper limb paresthesia and exhaustible bilateral plantar clonus and unstable gait.

Because CM-I does not present clear clinical signs, it is usually only diagnosed in adulthood (SCHIJMAN et al., 2004; ROMERO-LUNA et al., 2022). Symptoms vary depending on the dysfunction of the cervical spinal cord, compression of the brainstem and/or cerebellum (PAUL et al., 1983; HALPIN et al., 1990). The most common symptoms are occipital headache triggered by cervical movements or Valsalva maneuvers (a common clinical sign in 70-90% of patients), neuropathic pain in the cervical segments (40-70%), hyperreflexia in the lower limbs (51%), hand atrophy (35%), paresis in the upper limbs (33%) and lower limbs (17%). MC-I is associated with syringomyelia in 50-80% of cases (LÓPEZ et al., 2007).

Stovner (1993) investigated the headache associated with CM-I and defined it as similar to cervicogenic headache, almost always accompanied by dizziness. He suggested intracranial hypertension, compression of the brainstem and degeneration of the central part of the spinal cord as pathophysiological hypotheses.

Paul et al. (1983) reviewed 71 cases of CM-I and found that the most common clinical manifestations were pain in 69% of patients, muscle weakness in 56%, paresthesia/sensory deficit in 52% and imbalance in 40%. The signs of the neurological physical examination defined a foramen magnum compression syndrome in 22% of cases, central medullary syndrome in 65% and cerebellar syndrome in 11%.

In 2022, the patient underwent a CT scan of the skull which revealed fibrosis in the nape of the neck, and she was asked to correlate it with the clinical data for confirmation. The 2023 examination revealed no pathological calcifications.

As for the dura mater, there is no consensus on the most suitable procedure. Some authors recommend not violating it, others do (KRIEGER et al., 1999), and there are even those who advocate enlarging it by using a graft, a procedure called duroplasty, as they argue that this procedure is essential to prevent the formation of local fibrosis and recurrence of the clinical condition (DYSTE et al., 1989; FELDSTEIN; CHOUDHRI, 1999; MUNSHI et al., 2000).

4 CONCLUSION

Chiari malformation type I (CM-I) is the most benign of the 4 types described by Chiari and has a varied clinical presentation. The advent of computerized imaging is fundamental to its diagnosis, and bone decompression of the craniovertebral junction is still the main therapeutic strategy in these cases.

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