Available online at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/radcr

Case Report

Marchiafava-Bignami disease with typical imaging findings: A case report ☆

Noura Boukricha, MD, Wadii Bnouhanna, PhD, Mounia Rahmani, PhD, Maria Benabdeljlil, PhD, Saadia Aidi, PhD*

Ibn Sina University Hospital Center, Neurology A and Neuropsychology department, Rabat, Morocco

ARTICLE INFO

Article history:
Received 10 August 2024
Revised 23 September 2024
Accepted 24 September 2024

Keywords:
Marchiafava-bignami disease
Corpus callosum
MRI
Alcoholism

ABSTRACT

Marchiafava-Bignami disease is a rare neurological condition characterized by necrosis and demyelination of the corpus callosum, typically associated with chronic alcoholism and/or malnutrition. The clinical manifestations of Marchiafava-Bignami disease are diverse and often nonspecific. Diagnosis of Marchiafava-Bignami disease relies on magnetic resonance imaging findings, which reveal significant and symmetrical involvement of the corpus callosum. We report the case of a 48-year-old man with chronic alcoholism who has been experiencing symptoms of confusion, stupor, difficulties in using and manipulating objects, and balance disorders for the past 10 days. Brain magnetic resonance imaging revealed diffuse and complete involvement of the corpus callosum, characteristic of a severe form of Marchiafava-Bignami disease.

Introduction

Marchiafava-Bignami disease (MBD) is a rare neurological complication characterized by necrosis and demyelination of the corpus callosum (CC) [1]. It is primarily linked to chronic alcoholism and/or malnutrition, frequently associated with a deficiency in the B vitamin complex, particularly thiamine [2].

The clinical manifestations of MBD are diverse and often nonspecific, including neuropsychiatric disorders, dysarthria, tetraparesis, astasia-abasia, seizures, impaired consciousness, and symptoms of interhemispheric disconnection [1,3].

The diagnosis of MBD typically relies on both the medical history and magnetic resonance imaging (MRI), which reveals prominent and symmetrical involvement of the CC, particularly its midbody, genu, and splenium, with or without extracallosal lesions [3].

We report the case of a patient presenting with clinical features and imaging findings characteristic of an acute and severe form of MBD.

Case report

A 48-year-old man, with a 25-year history of chronic alcoholism that escalated to excessive levels in the last 4 months (2 liters per day of red wine), coupled with severe malnutrition

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

* Corresponding author.
E-mail address: noura.boukricha@gmail.com (N. Boukricha).

https://doi.org/10.1016/j.radcr.2024.09.132

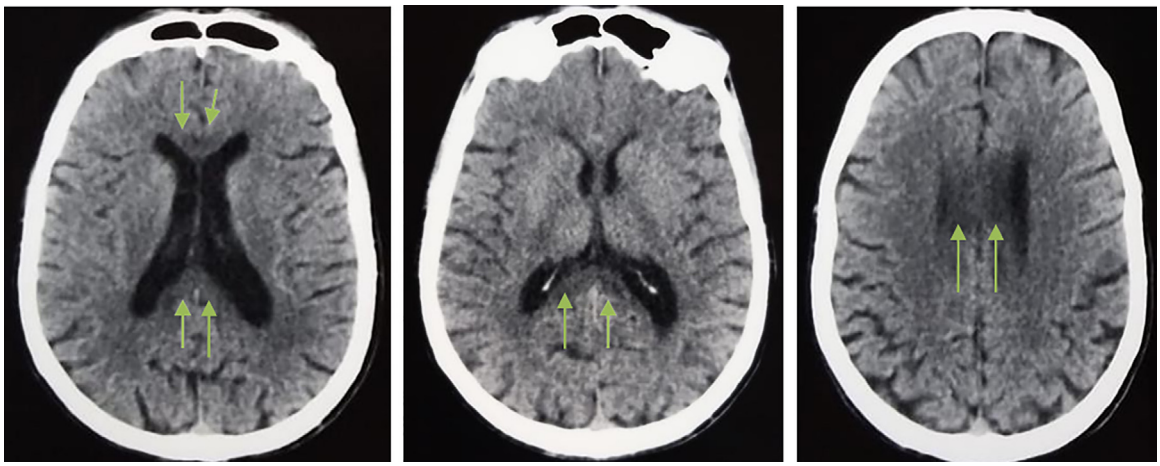

Fig. 1 – Brain CT scan showing hypodensity of the corpus callosum (green arrows).

over the past year, presented to our department with confusion, stupor, apathy, difficulties in using and manipulating objects, language and balance disturbances, and urinary incontinence, all of which had been evolving over the past 10 days.

The clinical examination revealed a cachectic and apathetic patient with dysarthria and comprehension difficulties. The patient also exhibited astasia-abasia, spastic hypertonia with hyperreflexia in all 4 limbs, gestural perseveration, and a bilateral grasp reflex.

The brain computed tomography (CT) scan showed hypodensity of the splenium of the CC (Fig. 1). The brain MRI, performed 2 days later, revealed hyperintensity on T2 and fluid-attenuated inversion recovery (FLAIR), and isosignal on T1 throughout the CC. Diffusion-weighted images (DWI) also showed hyperintensity in these regions with relatively decreased apparent diffusion coefficient (ADC) values, along with bilateral FLAIR hyperintensity in the frontal cortex (Fig. 2). The arterial MR angiography was normal (Fig. 3).

Laboratory studies revealed elevated serum gamma glutamyl transpeptidase, alanine transaminase and aspartate aminotransferase levels, a macrocytosis, and a decreased serum Vitamin B9.

The diagnosis of acute and severe MBD was established based on the patient's history, clinical manifestations, and MRI findings. The oral supplementation with a vitamin B complex (B1, B6, B12) was started due to the unavailability of the injectable form. The patient's condition worsened, leading to altered consciousness that required transfer to the intensive care unit and intubation. The patient died a few days later.

Discussion

The pathophysiological mechanism of MBD remains uncertain; however, published case reports regularly show that thiamine deficiency contributes to the development of the disease [3]. Cytotoxic edema appears to play a key role in the early stages, followed by demyelination and necrosis in the later stages [4,5]. Additionally, laminar sclerosis of the cerebral cortex, known as Morel's laminar sclerosis, is observed [6].

The symptoms of MBD can present acutely, subacutely, or chronically. The acute form is characterized by severe consciousness disturbances, seizures, and limb hypertonia. The subacute form presents with confusion, dysarthria, behavioral changes, drowsiness, and vision disorders. If diagnosis and treatment are not administered promptly, MBD can progress to coma or even result in death [2,7]. The chronic form, which is less common, typically manifests as persistent dementia [8].

Given the variability and nonspecific nature of the clinical presentation of MBD, early diagnosis and differentiation from other pathologies can be challenging. Nevertheless, MRI plays a key role in early diagnosis, as identifying pathognomonic signs is crucial for prompt management. The acute form of MBD is typically characterized by hyperintensity on T2/FLAIR and DWI sequences, hypointensity on ADC and T1, and swelling of the CC. The lesions observed are symmetric and may exhibit peripheral contrast enhancement. The observed lesions are symmetric and may show peripheral contrast enhancement [2].

A characteristic feature of acute MBD is the "sandwich sign," where the central body of the CC appears hyperintense on T2 and FLAIR MRI sequences, with relative sparing of the ventral and dorsal margins. In addition to CC abnormalities, other brain regions—including the cerebral cortex, hemispheric white matter, middle cerebellar peduncles, and basal ganglia—may also be affected [9].

Bilateral and symmetrical lesions in the cerebral cortex, particularly in the frontal lobes, have been documented in several radiological reports. These cortical lesions appeared hyperintense on T2/FLAIR sequences and showed restricted diffusion, which could suggest cytotoxic edema, representing the acute phase of Morel's laminar sclerosis [9]. Similarly, Ménégon et al. suggested that restricted diffusion affecting the entire CC, along with cortical involvement, might be indicative of a poor prognosis, both for survival and cognitive recovery [5].

In some cases, following the acute phase, edema resolves, and the T2 signal in the CC normalizes, eventually progress-

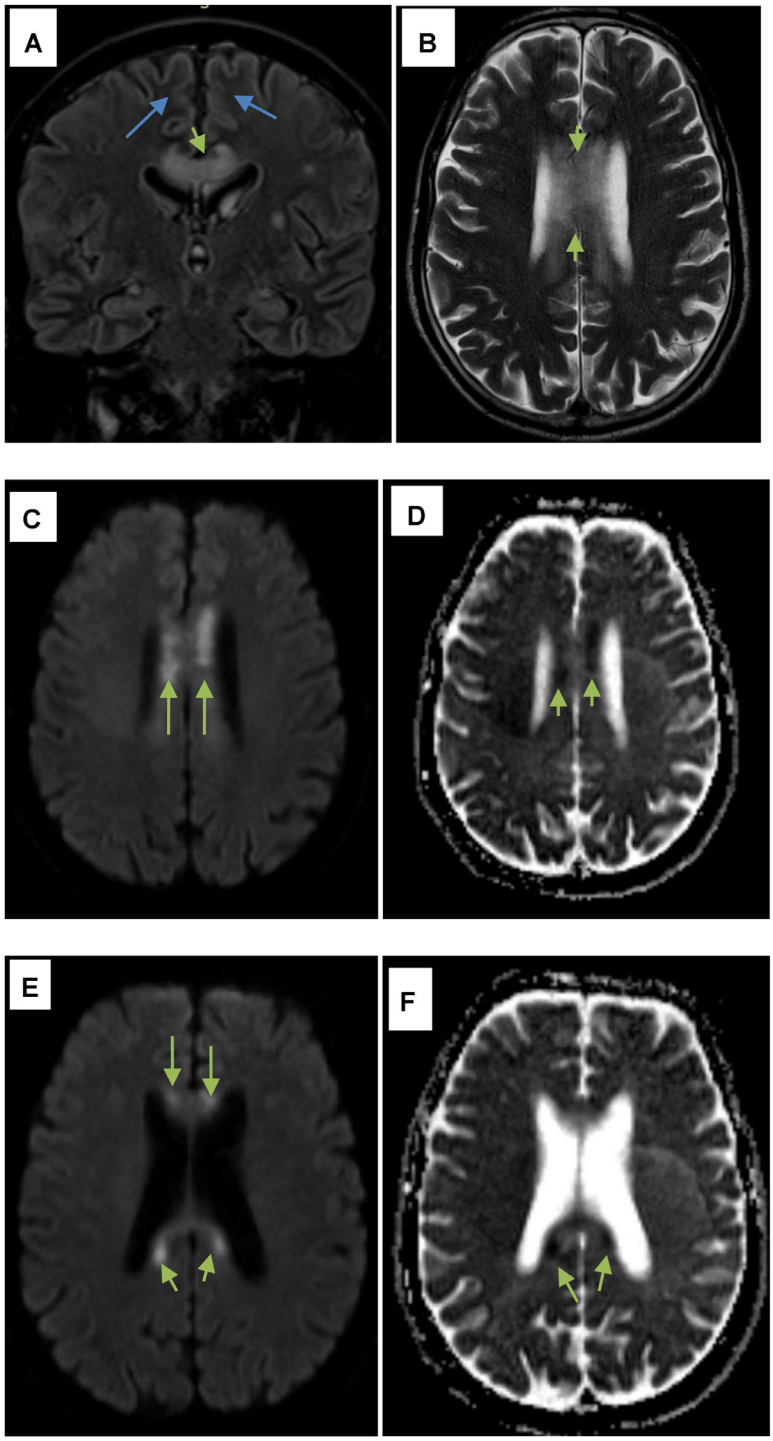

Fig. 2 – Brain MRI showing hyperintensity on FLAIR (A), T2 (B), and DWI (C, E) with relatively decreased ADC values (D, F) in the corpus callosum (green arrows), and bilateral FLAIR hyperintensity (A) in the frontal cortex (blue arrows).

ing towards symmetrical atrophy [10]. On CT, the CC typically shows hypoattenuating regions, although in cases of hemorrhage, these areas may appear iso- or hyperattenuating [9].

In 2004, Heinrich et al. proposed 2 subtypes of MBD based on clinical symptoms, imaging findings, and prognosis. Type A is characterized by the acute or subacute onset of major impairment of consciousness, diffuse edema involving the entire CC, and a poor outcome. Type B is characterized by a chronic course of cognitive disorders, signs of interhemispheric disconnection, partial lesions of the CC, and a favorable outcome [11]. Our patient is classified as type A due to the acute onset of severe symptoms, diffuse involvement of the entire CC, and the presence of extracallosal lesions noted in the frontal cortex on MRI.

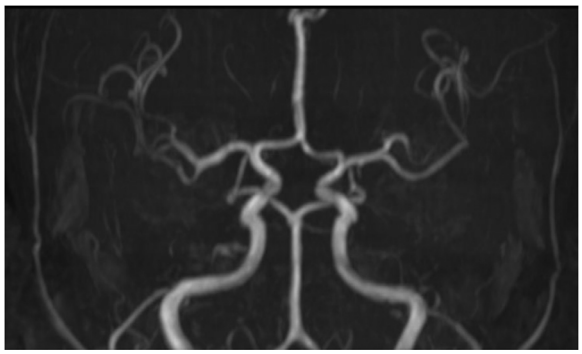

Fig. 3 – MR Angiography showing no abnormalities.

However, to establish an accurate differential diagnosis, it is essential to consider other conditions affecting the CC. It is particularly important to differentiate MBD from Wernicke's encephalopathy, which is also associated with alcoholism. The latter is characterized by involvement of the medial thalamic nuclei, hypothalamus, mammillary bodies, and periaqueductal gray matter. Pontine and extrapontine myelinolysis, on the other hand, is distinguished by specific involvement of the central pons, basal ganglia, thalamus, lateral geniculate body, cerebellum, and cerebral cortex. It is also necessary to differentiate MBD from other demyelinating diseases, such as multiple sclerosis (MS). In MS, involvement of the CC is often asymmetric and typically associated with other characteristic lesions, including periventricular, sub-tentorial, and spinal cord lesions. Finally, it is important to distinguish MBD from infarcts of the CC, which often present asymmetrically and are limited to a specific arterial territory. These infarcts can lead to focal lesions rather than diffuse involvement of the CC, which differentiates them from MBD [2,3].

High-dose parenteral thiamine and vitamin B complex supplementation remains the primary treatment for alcohol-related conditions and malnutrition. In addition to thiamine, steroids are also commonly used to treat MBD, as they can stabilize the blood-brain barrier and reduce inflammatory edema; however, their efficacy remains hypothetical [12,13].

The progression and prognosis of MBD can vary. Favorable outcomes with reversal of lesions on brain MRI are possible [14]. Severe consciousness disturbances, low ADC values of the CC, total involvement of the CC, and extracallosal lesions, like in our case, are factors that can be associated with a poor prognosis [1,10].

Conclusion

MBD is a rare condition that neurologists and neuroradiologists should consider when partial or diffuse damage to the CC, detected by MRI or CT, is observed in a patient with chronic alcoholism and malnutrition. Early diagnosis, combined with prompt treatment using parenteral thiamine, can offer the patient a chance of survival and recovery.

Patient consent

Informed written consent for the publication of this case was obtained from the patient's family. This consent includes permission to use the associated medical data, images, and relevant clinical details.

REFERENCES

[1] Shen YY, Zhou CG, Han N, Liang XM, Deng YQ. Clinical and neuroradiological features of 15 patients diagnosed with Marchiafava-Bignami disease. Chin Med J (Engl) 2019;132(15):1887–9.

[2] Dong X, Bai C, Nao J. Clinical and radiological features of Marchiafava-Bignami disease. Medicine (Baltimore) 2018;97(5):e9626.

[3] Kinsley S, Giovane RA, Daly S, Shulman D. Rare case of Marchiafava-Bignami disease due to thiamine deficiency and malnutrition. BMJ Case Rep 2020;13(12):e238187.

[4] Friese SA, Bitzer M, Freudenstein D, Voigt K, Küker W. Classification of acquired lesions of the corpus callosum with MRI. Neuroradiol 2000;42:795–802.

[5] Ménégon P, Sibon I, Pachai C, Orgogozo JM, Dousset V. MarchiafavaBignami disease: diffusion-weighted MRI in corpus callosum and cortical lesions. Neurology 2005;65:475–7.

[6] Kawarabuki K, Sakakibara T, Hirai M, Yoshioka Y, Yamamoto Y, Yamaki T. Marchiafava-Bignami disease: magnetic resonance imaging findings in corpus callosum and subcortical white matter. Eur J Radiol 2003;48:175–7.

[7] Kumar KS, Challam R, Singh WJ. Marchiafava–Bignami disease: a case report. J Clin Diagn Res 2014;8:RD01–2.

[8] Perea J, Luis MB, Lázaro LG, Scollo S, Tamargo A, Crespo J, et al. Marchiafava-Bignami disease associated with spinal involvement. Case Rep Neurol Med 2020;2020:8867383.

[9] Waack A, Nandwani S, Ranabothu M, Ranabothu A, Vattipally V. Marchiafava-Bignami disease: case presentation and radiological imaging. Radiol Case Rep 2023;18(11):3922–5.

[10] Wenz H, Eisele P, Artemis D, Förster A, Brockmann MA. Acute Marchiafava-Bignami disease with extensive diffusion restriction and early recovery: case report and review of the literature. J Neuroimaging 2014;24(4):421–4.

[11] Heinrich A, Runge U, Khaw AV. Clinicoradiologic subtypes of Marchiafava-Bignami disease. J Neurol 2004;251(9):1050–9.

[12] Hillbom M, Saloheimo P, Fujioka S, Wszolek ZK, Juvela S, Leone MA. Diagnosis and management of Marchiafava-Bignami disease: a review of CT/MRI confirmed cases. J Neurol Neurosurg Psychiatry 2014;85(2):168–73.

[13] Tao H, Kitagawa N, Kako Y, Yamanaka H, Ito K, Denda K, et al. A case of anorexia nervosa with Marchiafava-Bignami disease that responded to highdose intravenous corticosteroid administration. Psychiatry Res 2007;156(2):181–4.

[14] Muccio CF, De Lipsis L, Belmonte R, Cerase A. Reversible MR findings in Marchiafava-Bignami disease. Case Rep Neurol Med 2019;2019:1951030.