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Case Report

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Marchiafava–Bignami disease: A case report

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ABSTRACT

Rationale: Clinicians encounter multiple alcohol-related illnesses in practice, and Marchiafava-Bignami disease is a rare and devastating entity among them. It is a toxic-demyelinating disease and seen in chronic alcoholics, although it may be occasionally observed in chronically malnourished teetotalers. The clinical presentations are diverse. The symptoms and signs are non-specific, and the onset can be acute or chronic.

Patient's concerns: A 45-year-old right-handed patient suffered from alcohol use disorder with multiple non-specific neuropsychiatric manifestations.

Diagnosis: Marchiafava-Bignami disease.

Interventions: Thiamine, folate, vitamin B12, and steroid therapy.

Outcome: The patient's behaviour was significantly improved but dysarthria and pyramidal signs persisted. He was left with permanent cognitive impairment.

Lessons: Though prompt therapy may halt the demyelinating process in this disease, the treatment remains a challenge in clinical practice. The recognition of the neuro-radiologic features is crucial to establish an early diagnosis.

KEYWORDS: Marchiafava-Bignami disease; Alcohol use disorder; Magnetic resonance imaging

1. Introduction

Marchiafava-Bignami disease (MBD) is an alcoholism-related rare entity characterized by demyelination and necrosis of the corpus callosum and adjacent subcortical white matterradiologically. Despite the etiologically debatable condition, it is presumed to develop due to alcohol-induced neurotoxicity coupled with malnutrition, leading to cytotoxic edema, demyelination, metabolic and vascular disturbances[1,2]. The symptoms and signs are non-specific, and the onset can be acute or chronic. The neurological abnormalities include cognitive impairment, ataxia, dysarthria, depression, seizures, pyramidal defects leading to spasticity and hyperreflexia, consciousness impairment which can be severe enough to render the patient comatose. Split-brain or callosal disconnection syndrome can depend on the fibres affected, which leads to a multitude of manifestations such as aphasia, apraxia, prosopagnosia, hemiparesis, and locked-in-syndrome[3].

2. Case report

This study was supported by the Ethical Committee of Government Medical College Hospital. Informed consent has been obtained from the patient.

A 45-year-old right-handed, underweight, and incontinent male was referred to our hospital in a stuporous and hemodynamically stable state. Collateral history revealed that he had been suffering from alcohol use disorder as he consumed more than 100 g of country-made liquor for the last 3 years. There was no obvious precipitant for the current illness. The symptoms were progressive and first appeared just over a week back with the symptoms of low mood, agitation, and dysarthria, followed by obtundation. There was no history of seizures, fever, vomiting, headache, head injury, hypertension, diabetes mellitus, tuberculosis, or any other chronic illness. Previous medical history did not suggest any blood transfusions, needle prick injury, or promiscuous behavior.

Physical examination revealed that the patient was malnourished with altered sensorium. The patient had a pulse rate of 90/min and blood pressure of 110/80 mm of Hg. There was no pallor, icterus,

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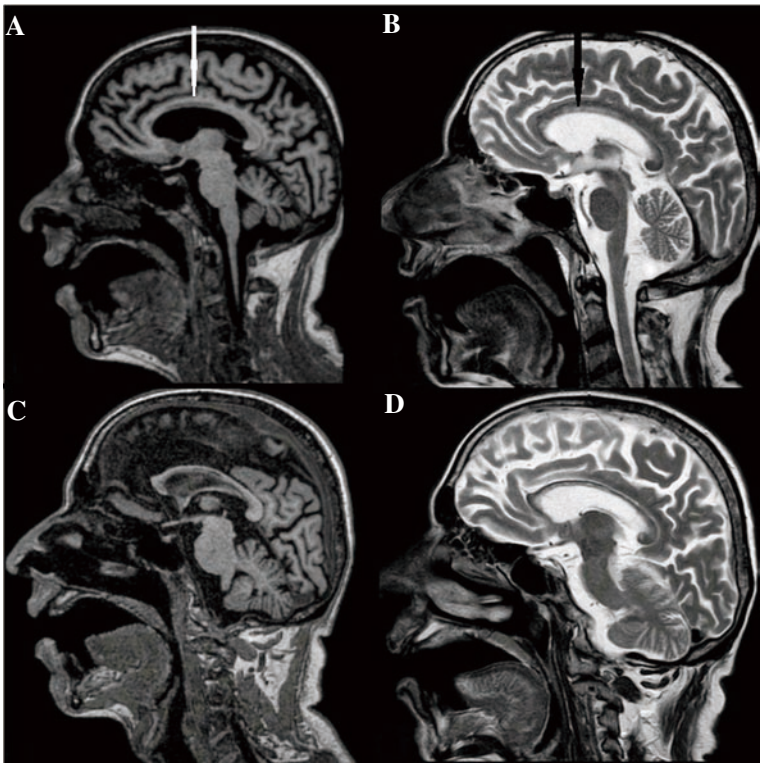

Figure 1. Brain MRI of a 45-year old man with Marchiafava-Bignami disease before and after treatment. A: Pre-treatment sagittal T1 weighted MRI reveals areas of necrosis in the body of the corpus callosum (white arrow). B: Pre-treatment Sagittal T2 weighted MRI reveals hyperintense signal involving the corpus callosum (black arrow). C and D: Post-treatment Sagittal T1 and T2 weighted MRI reveals no radiological improvement.

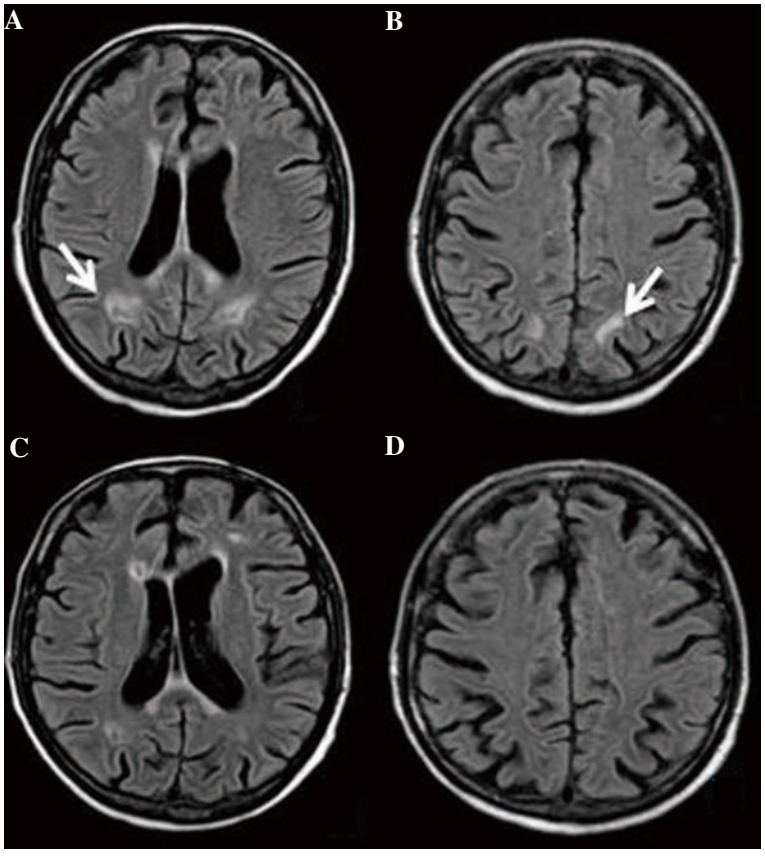

Figure 2. Pre and post-treatment axial fluid-attenuated inversion recovery MRI. A and B: Pre-treatment axial fluid-attenuated inversion recovery MRI reveals altered signal intensity in the parietal subcortical white matter (white arrows). C and D: Post-treatment axial fluid-attenuated inversion recovery MRI reveals partial resolution of subcortical white matter abnormality.

cyanosis, clubbing, and lymphadenopathy presented in the patient. On detailed neurological examination, he had scarce spontaneous movements with minimal verbal and motor response to even painful stimuli. The limbs were spastic with hyperreflexia, and the pupils reacted sluggishly. No nystagmus or meningeal signs were notable on examination. The fundus examination was normal. There was no evidence of involuntary movements. Respiratory examination revealed vesicular sounds over both lungs without any added sounds. Cardiovascular examination was unremarkable.

After a panel of investigations including magnetic resonance imaging (MRI) of the brain (Figure 1) and normal cerebrospinal fluid examination, diagnosis of MBD was made. Blood investigations revealed macrocytic anemia (hemoglobin: 11.2 g/dL, normal range:14-18 g/dL), and mildly elevated hepatic transaminases and gamma-glutamyl transferase. Viral markers, prothrombin time, and serum ammonia levels were normal. Chest X-ray and ECG were normal. Blood and urine cultures were sterile. Ultrasonography of the abdomen showed fatty liver with mild hepatomegaly without evidence of portal hypertension.

The patient was administrated with intravenous thiamine (300 mg/d for 5 d), vitamin B complex, vitamin B12 (1 000 μg/d for 5 d), folate (5 mg/d) from day 1 of admission. However, the sensorium of the patient remained the same. So, on day 3 he was initiated on 1000 mg of intravenous methylprednisolone which was continued for 3 d. However, his response was suboptimal as he continued to be drowsy and on nasogastric support at 2 weeks.

On discharge, the patient was advised strict alcohol abstinence with rehabilitation and proper nutrition. After 6 weeks of treatment, though there was a significant improvement in his behaviour but dysarthria, pyramidal signs persisted. He was left with permanent cognitive impairment as assessed by the Mini-Mental Scale examination score (22/30, normal range: 25 or above 25). However, there was definite resolution in the radiological lesions on repeat MRI (Figure 2).

3. Discussion

MBD is a rarely encountered entity. Though characterized by variable symptomatology, it often presents with psychotic and emotional symptoms, seizures and hemiparesis, reduced consciousness or coma, possibly culminating in death. The extent of radiological damage correlates with the clinical picture and outcome, and rarely spinal cord may also be involved[4]. Callosal atrophy itself, without overt clinical manifestations, is very common in alcoholics[3]. The radiological lesions, in the form of T2/fluid-attenuated inversion recovery sequence hyperintensities on MRI, first appear in the body with delayed involvement of the genu and splenium of the corpus callosum[5]. Chronic forms can display callosal thinning, atrophy, and cystic changes as well as concurrent cerebral atrophy. In the absence of cystic lesions, some degree of radiographic reversal is possible[6]. Lesions outside the corpus callosum are uncommon, though some studies have documented simultaneous lesions in frontal, periventricular white matter, and middle cerebellar peduncles[5].

The differential diagnoses comprise of clinical or radiologic mimics such as stroke, encephalitis, multiple sclerosis, lymphoma, and Alzheimer's disease, and other neurologic manifestations related to alcohol use such as Wernicke encephalopathy, Korsakoff, and osmotic demyelination syndrome, hypoglycemic or hepatic encephalopathy, and delirium tremens.

Besides, mandatory cessation of alcohol intake, therapeutic dosages of thiamine, folate, and vitamin B complex, amantadine, as well as high-dose corticosteroids have been attempted in various case reports[7,8]. These treatment options are based on the proposed etiopathogenic hypothesis of deficiency and the observed response with therapeutic doses of vitamins. Corticosteroids are believed to stabilize the blood-brain barrier, diminish inflammatory cell cascade, and resulting cytotoxic edema. It is important to realize that majority of MBD are treated with both steroids and multivitamins simultaneously, and the individual effect of each cannot be confirmed. The recovery is however never complete and survivors can be left with crippling dementia. Recovery depends upon the extent of extra-callosal lesions and cerebral grey and white matter involvement, although diagnostic suspicion and prompt empirical management are just as essential[9,10].

The underlying pathogenesis of MBD is still not clearly understood. A multitude of metabolic, toxic, and vascular disturbances interact with malnutrition and chronic alcoholism. Due to uncertainty in its etiology, specific therapy is also not available. Besides cessation of alcohol, high doses of corticosteroids and vitamin B complex, including thiamine, vitamin B12, and folate have been therapeutically utilized as in our case. The outcome of these therapies is variable.

Conflict of interest statement

The authors report no conflict of interest.

Authors' contributions

All the authors have provided substantial contributions in the clinical management of the case and literature review on the topic in question. S.G. (Swati Garg) and S.G. (Saurabh Gaba) have drafted the manuscript, reviewed the literature and M.G. (Monica Gupta) and R.G. (Rekha Gupta) have analysed and revised it critically for important intellectual content. All the authors have read the final version and approved it. All the authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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