Clinical Presentation and Radiological Findings in Marchiafava-Bignami Disease

Dear Sirs,

Marchiafava-Bignami disease (MBD) is a rare disorder of unknown etiology. Alcoholism is the greatest risk factor for MBD. The disease is characterized by progressive symmetrical demyelination and necrosis of corpus callosum. The clinical features of MBD are non-specific and it presents with a plethora of neuropsychiatric symptoms.[1] The modern diagnosis is almost always based entirely on the magnetic resonance imaging (MRI). There are several indications that this disease may be underdiagnosed. Early diagnosis and treatment of the disease leads to a favorable outcome.[2] We report a case of MBD in a middle-aged man with chronic alcohol abuse who presented with a cluster of neuropsychiatric symptoms.

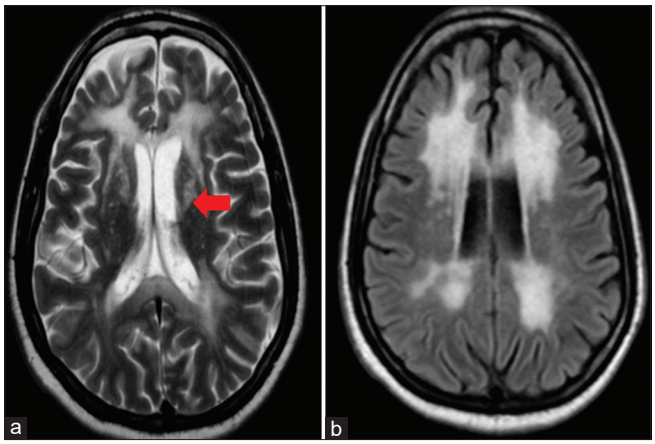

Figure 1: Imaging MRI of the case report (a) T2 AXIAL: Confluent symmetrical hyperintensities noted in the periventricular and subcortical white matter of bilateral frontal and parietal lobe, T2 hyperintensity is noted involving anterior limb of internal capsule (Red arrow) and anterior part of corpus callosum; (b) FLAIR AXIAL image showing confluent hyperintensities in the periventricular region. Source: Author's own work

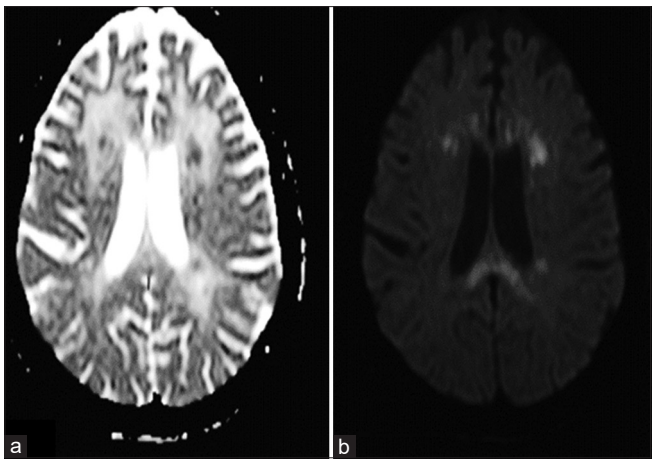

Figure 3: Imaging MRI of the case report (a) Apparent diffusion coefficient (ADC) showing areas of reduced signal intensity in bilateral periventricular white matter; (b) - Diffusion weighted image (DWI) showing high signal intensity areas in bilateral periventricular white matter and splenium of corpus callosum. Source: Author's own work

A 36-year-old gentleman with seven years history of alcohol dependence presented to the psychiatry outpatient department with three months history of behavioral change. There was a history of slowness of all activities, poor oral intake and standing in the same place for few minutes at times. In addition, there was a marked reduction in interaction with family members, slurred incomprehensible speech, and emotional lability. He required assistance in performing all his activities of daily living and occasions of fecal and urinary incontinence were also reported. There were no withdrawal symptoms. Mental status

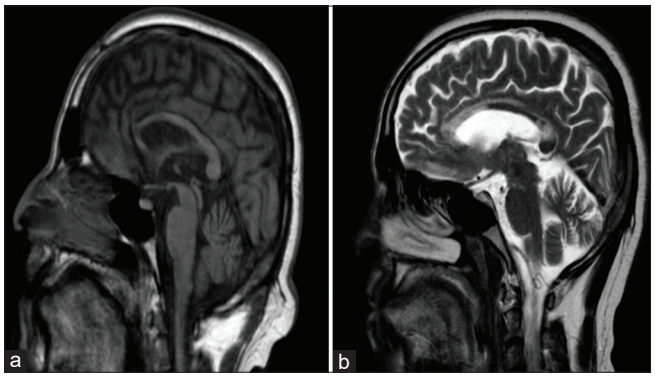

Figure 2: Imaging MRI of the case report (a) Para sagittal T1 W MRI – Thinned out corpus callosum. Linear hypointense areas noted involving the central layers of corpus callosum; (b) T2-SAGITTAL MRI – Hyper intensities noted involving the rostrum, genu, anterior body and splenium of corpus callosum without any mass effect. Source: Author's own work

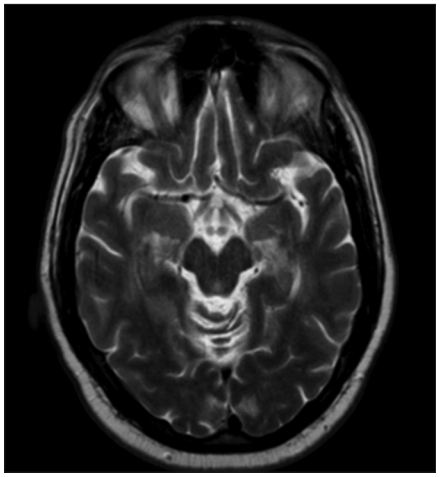

Figure 4: Bilateral normal mammillary bodies. Source: Author's own work

examination revealed retardation in psychomotor activity, mask like facies, near mutism, occasional incomprehensible speech, and perseveration. His neurological examination revealed unsteady gait and global aphasia, no abnormalities were present in motor and sensory system examination and there were no cerebellar signs. Ocular examination revealed vertical nystagmus to downward gaze and fundus examination was normal. Hindi Mental State Examination (HMSE) score at the time of admission was 9/31, indicative of severe cognitive impairment. Electroencephalogram (EEG) showed bilateral hemispherical dysfunction and Computed tomography (CT) brain revealed small vessel ischemic changes and cerebral atrophy. Subsequent MRI brain revealed T2 hyperintensity involving the periventricular and subcortical white matter of bilateral frontal and parietal lobe, the genu, rostrum, and splenium of corpus callosum, with thinning of genu and rostrum [Figures 1-4]. The diagnosis of MBD was made based on the clinical presentation of neuropsychiatric symptoms and MRI findings of symmetrical corpus callosal involvement. He was started on parenteral thiamine supplementation up to 1500 mg per day in divided doses and oral lorazepam 2 mg was given at the night for insomnia. Patient was followed up for a period of three months and there was a partial improvement in his cognition and behavioral disturbance. The HMSE score improved to 18/31 from a baseline score of 9/31. He was continued on oral thiamine supplementation.

The diagnosis of MBD was suspected in this patient with history of alcohol dependence who presented with behavioral symptoms along with plethora of neuropsychiatric symptoms such as emotional lability, slowness of movements, gait disturbances, global aphasia, memory disturbances along with fecal and urinary incontinence and MRI imaging confirmed the diagnosis of MBD based on symmetrical corpus callosal involvement, especially seen with T2 weighted images showing hyperintensities of genu, rostrum, and splenium of corpus callosum with thinning of genu and rostrum. Supplementation with parenteral thiamine during hospitalization and later with oral thiamine supplementation led to partial improvement in his cognition and behavioral symptoms at the end of three months follow-up.

MBD was first described in1903 by the Italian pathologists Amico Bignami and Ettore Marchiafava in three Italian patients with a history of red wine consumption. MBD is rare disease and does not have typical clinical presentation as it is difficult to differentiate from other diseases at an early stage.[3] The corpus callosum appears hypo attenuated on CT scans, with the exception of cases that are characterized by subacute bleeding, in which it may be iso to hyper attenuated. Conventional MRI typically detects lesions as hyperintense on T2- phase and FLAIR signal intensity, and hypointense on T1-weightedimages in the body of the corpus callosum, sometimes extending into the genu, and the splenium.[4] Early diagnosis of MBD is typical lesions limited to genu, body and splenium of corpus callosum. It can also affect bilateral cerebral hemispheric white matter and basal ganglia. Presence of areas of diffusion restriction within the involved cortical areas could represent the cytotoxic edema which indicates acute phases of Morel's laminar cortical necrosis.[5]

Early stage is characterized by diffuse swelling of the corpus callosum in the form of T1 hypo intensity and T2 hyperintensity of corpus callosum. After the acute stage, edematous change subsides and the corpus callosum recovers its normal intensity. In chronic stage, it is characterized by diffuse atrophy of corpus callosum with areas of corpus callosum that appear as T1 hypointense and T2 hyperintense areas confined to genu, body, and splenium of corpus callosum. CT is not useful in diagnosis, while MRI is better in diagnosing and follow-up.[6]

In an alcoholic with abovementioned spectrum of imaging findings, the differentials would be MBD and Wernicke's encephalopathy. In Wernicke's encephalopathy, the mammillary bodies will be atrophied and show high signal intensity on T2 weighted images. Also, there will be T2 hyperintensity along the medial thalamus, periaqueductal grey matter.[7,8]

There is no specific treatment regimen approved for MBD.[9] However, studies have proved that early initiation of treatment with parenteral thiamine has demonstrated a statistically significant outcome when compared with delayed treatment and has improved the prognosis of MBD from frequently fatal to a mortality of less than 8%.[10] The early initiation of treatment within two weeks of symptom onset is the most important predictor of treatment outcome in those treated with thiamine when compared with those with delayed treatment initiation. Although steroids had been tried in treating MBD, it lacks significant results like those observed with treatment with parenteral thiamine. Further studies are required to establish a standard treatment regimen and for better understanding about the disease.

MBD should be considered in patients with alcohol abuse who present with neuropsychiatric symptoms. A high index of suspicion and an early MRI in such patients can facilitate early detection and treatment of MBD. Typical lesions of the corpus callosum can aid in the diagnosis and an early initiation of treatment with parenteral thiamine and multivitamins can facilitate a favorable outcome.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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Submitted: 20-Mar-2022 **Revised:** 19-May-2022 **Accepted:** 21-May-2022
Published: 31-Oct-2022

DOI: 10.4103/aian.aian_267_22