Marchiafava-Bignami Disease: A Rare Clinical Dilemma

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Abstract

Marchiafava-Bignami Disease (MBD) is a progressive neurological disease, characterized by corpus callosal demyelination and necrosis and subsequent atrophy. It is usually seen in the context of alcoholism and malnutrition. Clinical diagnosis of this disease is quite challenging due to various presentations but a high degree of suspicion often leads to the correct diagnosis with help of neuroimaging. We report a case of MBD with a classical clinical course and typical radiological features. This case is highlighted in order to generate awareness regarding this uncommon but historic complication of chronic alcoholism.

Introduction

Marchiafava-Bignami disease (MBD) is a progressive neurological disease, characterized by corpus callosal demyelination and necrosis and subsequent atrophy. It is usually seen in the context of alcoholism and malnutrition. In 1903, Italian pathologists Marchiafava and Bignami described 3 alcoholic men who died after having seizures and coma. In each patient, the middle two thirds of the corpus callosum was found to be severely necrotic. It can pose diagnostic dilemma in an appropriate scenario (such as heavy alcohol consumers/malnourished individuals) by mimicking the other common aetiologies of dementia, seizures, behavior disturbances and gait abnormalities.

MBD is a very rare condition. In 2001, Helenius et al.¹ wrote that they had found approximately 250 cases in published reports, although they also suggested that many cases had gone undiagnosed.

Case Report

A 40 years old healthy male was admitted to our medical ward with complaints of insidious onset gradually progressive gait instability for past 1 year and numbness of both lower limbs for past 15 days. There was no history of seizures, bladder-bowel-bladder disturbances or any history suggestive of cranial nerves involvement. There was no history of trauma in past. He had been a chronic alcoholic since last 15 years and he used to have 350-400 ml of country liquor daily. He was conscious, oriented to time, place and person but aggressive and irritable during the examination. He had severe spasticity of all four limbs with normal power. All deep tendon reflexes were exaggerated and bilateral plantar reflexes were flexor. He also had signs suggestive of cerebellar involvement like dysarthria and impaired coordination. He had sensory involvement in the form of reduced vibration and joint position sense. Gait was wide based and both spastic and ataxic type. There were no signs of inter-hemispheric callosal disconnection. Rest of the systemic examination was normal. On further laboratory evaluation he had a normal complete blood count with peripheral blood film. His liver function tests were deranged i.e. total bilirubin was 3.6 mg/dl, direct bilirubin was 1.9 mg/dl. Aspartate aminotransferase (AST or SGOT) was 275 W/L, alanine aminotransferase (ALT or SGPT) was 64 W/L, alkaline phosphatase (ALP) was 209 W/L, total protein was 7.9 gm/dl, albumin was 2.9 g/dl indicating albumin and globulin ratio reversal. His renal function tests, blood sugar, serum electrolytes were found to be within normal limits. He had normal thyroid profile with normal vitamin B12 levels. Blood HIV and VDRL tests were negative. His chest x-ray and electrocardiogram was normal. Ultrasonography revealed enlarged fatty liver with altered echogenicity without any evidence of portal hypertension. Magnetic resonance imaging (MRI) of brain with cervical spine was performed which revealed thinning of corpus callosum with abnormal signals within it, periventricular white matter and corona radiata, associated with changes of diffuse cerebral and cerebellar atrophy with normal cervical spinal cord (Figure 1). Based on the clinical scenario of the patient and neuroimaging findings a diagnosis of Marchiafava-Bignami disease was made. Patient was provided supportive treatment for his illness and advised complete abstinence from alcohol.

Discussion

Specific clinical characteristics of this case are chronic alcoholism, progressive gait imbalance, behavioral disturbances, dysarthria, spasticity, sensory involvement, deranged liver function tests and MRI findings of thinning of corpus callosum with diffuse cerebral and cerebellar atrophy.

MBD is most frequently seen in middle-aged or elderly chronic alcoholic males.¹ MBD was first reported in 1903 by Marchiafava and Bignami, who originally described the symptoms in Italian men with increased consumption of inexpensively manufactured Chianti red wine.¹ Currently, however, MBD is known to occur in patients with chronic consumption of other sorts of alcohol including whisky and French liqueur.¹ MBD has also been found in severely malnourished people without a history of alcoholism.¹ In the present case, long-term consumption of alcohol in form of country liquor might have been related to the pathogenesis of MBD. Although the precise mechanisms underlying development of MBD remain unknown, effects of toxic agents present in alcohol, vitamin-B complex deficiency, or osmotic

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disorders have been considered as potential causes. The clinical spectrum of this disease is diverse which makes diagnosis difficult. In acute cases, prognosis is usually poor and mortality is high, even though there are case reports documenting complete recovery. Acute MBD and Wernicke’s encephalopathy clinically may occur together. Differentiating acute MBD from Wernicke’s encephalopathy is not difficult, because in the latter, MR imaging shows abnormal signal intensity and contrast enhancement in the mammillary bodies, periaqueductal region, and the walls of the third ventricle. Chronic presentation includes progressive dementia, behavioural abnormalities and an interhemispheric disconnection syndrome (limb apraxia, tactile agraphia, unilateral agraphia and hemialexia). An intermediate form has been described with an initial acute onset followed by regression to chronic form. It may be seen in combination with other manifestations of chronic alcohol abuse like Wernicke’s encephalopathy, Central Pontine myelinolysis and Morel’s laminar sclerosis. Our patient had signs of corticospinal tract, cerebellar and posterior column involvement simulating either subacute combined degeneration of cord or spinocerebellar ataxia with alcoholic neuropathy, however these two differentials were ruled out following laboratory investigations and MRI brain findings. In the past, cases of MBD were diagnosed only at autopsy, but with the advent of CT and MRI of brain findings. In the past, cases of MBD were diagnosed only at autopsy, but with the advent of CT and MRI of brain, early and prompt diagnosis has resulted in improved survival and better prognosis. The characteristic MRI picture of acute MBD shows symmetrical lesions involving the central portion of the body of corpus callosum with sparing of dorsal and ventral layer; the ‘sandwich sign’. Sometimes, lesion extends into the genu and splenium, but only rarely is the entire corpus callosum involved. The lesions are hypointense on T1WI, hyperintense on T2WI and FLAIR showing diffusion restriction on DWI and variable reduction in apparent diffusion coefficient value. Recent MRI studies have shown that lesions may also be found in the cerebral hemispheric white matter, cerebellar peduncles or cortical grey matter. Our patient had typical lesions involving corpus callosum and additional extracallosal lesions involving periventricular white matter and corona radiata associated with changes of diffuse cerebral and cerebellar atrophy. In 2004, Heinrich et al. described 2 clinical subtypes of MBD as follows:

- **Type A** - This has predominant features of coma and stupor; this subtype is associated with a high prevalence of pyramidal-tract symptoms; radiologic features include involvement of the entire corpus callosum.

- **Type B** - Characterized by normal or mildly impaired mental status; radiologic features are partial or focal callosal lesions.

Our patient is categorised into Heinrich type A in view of the clinicoradiological picture. There is no specific therapy for MBD. Prompt diagnosis and early initiation of treatment with thiamine, vitamin B complex and folic acid expedite clinical recovery. High-dose steroids may aid in recovery by reducing oedema.

### Conclusion

MBD is a disorder found in chronic alcoholics and malnourished patients. It should be included in differential diagnosis of an alcoholic patient presenting with neurological manifestations. MBD can mimic various neurological diseases and should be ruled out by neuroimaging of brain, which is a key to the diagnosis. Alcohol abstinence along with thiamine, vitamin B complex with vitamin B12 and folic acid can be given for treatment.

### References


