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Marchiafava Bignami Disease – Typical and Atypical Etiologies

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Abstract*—Marchiafava Bignami Disease (MBD) is a rare toxic demyelinating neuroetiology conventionally associated with chronic alcoholism. It can also be seen in patients with chronic malnutrition. The presentation is with neurocognitive manifestations. Typical involvement of corpus callosum on MRI is what clinches the diagnosis. MRI with diffusion weighted imaging is imperative in early disease for timely intervention and prognostication. Lucid description of two cases is presented – with typical and atypical etiologies – followed by detailed discussion of the disorder.*

***Keywords*—** *Demyelinating, corpus callosum, malnutrition, diffusion weighted MRI.*

I. INTRODUCTION

Marchiafava Bignami Disease (MBD) is a rare toxic disease characterized by progressive demyelination and necrosis of corpus callosum. It is seen mostly in male chronic alcoholics; however, we encountered features of acute MBD in a female patient with severe malnutrition. Further description entails two patients of MBD – with typical and atypical etiologies – followed by detailed discussion of the disorder.

Patient with Typical Etiology

58 year old male patient, chronic alcoholic with hepatic cirrhosis, under evaluation in psychiatry department for harmful use of alcohol, presented in emergency OPD with acute onset of seizures and two days history of Parkinsonian symptoms. On examination, patient was conscious, oriented, GCS was normal. Cog wheel rigidity was noted. Patient was moving all four limbs. Detailed lab investigations revealed thiamine and albumin deficiency and hypoglycaemia. MRI brain revealed T2WIand FLAIR hyperintensities in body of corpus callosum and bilateral subcortical white matter. The lesions did not show restriction of diffusion. Patient was managed with intravenous thiamine, glucose supplementation. He was discharged with advice on alcohol cessation and diet modification.

Patient with Atypical Etiology

A 36 year old female, mother of two children aged 16 and 12 years, presented to routine medical OPD with complaints of blurring of vision, diplopia and tinnitus, off and on for past 3-4 months. There was history of significant weight loss 30-34 kgs over past 6-7 months (Patient gave history of religious belief that her fasting will please the God and her son will perform well in academics). On examination, patient was a malnourished individual with short and long term memory deficit. There was agraphia, ataxia, wide based gait and rigidity. Bilateral horizontal gaze palsy, upbeat nystagmus on vertical gaze, Romberg sign positive, plantar reflexes directed downwards were noted. No obvious cerebellar signs were noted. On this basis, a differential diagnosis of 1) brainstem lesion 2) Nutritional VitB12 deficiency 3) Paraneoplastic syndrome was entertained. Lab investigations revealed severe thiamine, Vit B12 and albumin deficiency. NCCT head revealed normal study. MRI brain revealed no lesion on T1 and T2WI. On Diffusion weighted images, there was focal lesion in the splenium of corpus callosum with true restriction of diffusion. Similar lesions were also noted in bilateral periventricular and subcortical white matter. On this basis, a final diagnosis of MBD was made and patient was administered intravenous thiamine. On a follow up MRI brain after 3 months, the lesions had decreased in number and at 6 months, the lesions were imperceptible with no recurrence of visual symptoms in the patient. She is still under follow up at psychiatry OPD for evaluation of anorexia.

Discussion

MBD is rare demyelinating neurological disorder conventionally associated with chronic alcohol consumption and rarely seen in non-alcoholic patients (as occurred in atypical etiology case). The causative agent for this disease is known to be deficiency of VitB12/B complex. Typical diagnostic lesions occur in corpus callosum which are easily identifiable on MRI. The patients show improvement after administration of intravenous thiamine.

History

MBD in an era was primarily occurring in central regions of Italy where people consumed large quantities of scrupulously manufactured red wine. However, in recent times, MBD is known to occur worldwide and all alcoholic beverages are now implicated. Alcohol per se is not the causative agent, but chronic alcoholism and addiction leads to preferential intake of alcohol over food and eventually deficiency of thiamine and Vit B12 causing MBD.

Clinical Features

The typical patient is a male 40-60 years of age, chronic alcoholic. However, it can also be noted in cases of chronic malnutrition as was noted in the atypical etiology case. The onset of disease can be acute, subacute or chronic. Acute

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presentation is with altered sensorium, seizures and limb hypertonia. Subacute form presents with confusion, dysarthria, behavioural abnormalities and visual symptoms. Chronic presentation is in the form of dementia.

MRI Findings

Typical findings are bisymmetric lesions of corpus callosum, mostly involving the splenium. Early feature on MRI includes areas of diffusion restriction in corpus callosum, especially in splenium. This occurs due to cytotoxic edema. This is followed by T2WI/FLAIR hyperintensities in corpus callosum. Lesions may also occur in subcortical white matter and white matter tracts like anterior and posterior commissure. Lesions, if seen in extracallosal sites, predict a poorer outcome and severely impaired cognition on follow up. In chronic cases, the corpus callosum undergoes laminar necrosis with thinning and formation of cystic cavities.

Differential Diagnosis

Acute MBD needs to be differentiated from two common disorders.

1) Wernicke's encephalopathy – this is another alcohol related disease with acute presentation. Presenting symptoms are ataxia, ophthalmoplegia, nystagmus and confusion. MRI brain shows T2WI/FLAIR hyperintensities in medial thalamic nuclei, hypothalamus, mammillary bodies and periaqueductal gray matter.
2) Pontine and extrapontine myelinolysis – In these cases, MRI brain shows involvement of central pons, basal ganglia, thalami, lateral geniculate body, cerebellum and cerebral cortex.

Chronic MBD needs to be differentiated from other forms of chronic dementias like Alzheimer's disease, multi infarct dementia, Niemann- Pick disease, Korsakoff psychosis.

Diagnosis

Since MBD is still reported to be a rare disease, does not have a typical presentation, hence, it is difficult to diagnose and differentiate from other disorders. The diagnosis is a clinic- patho- radiological correlate. High degree of suspicion is warranted in a chronic alcoholic /malnourished patient presenting with neurological symptoms. Decreased levels of thiamine and callosal lesions on MRI clinche the diagnosis.

Treatment

Since the causative agent is deficiency of VitB12/B complex, intravenous administration of loading doses of thiamine relieves the patient of deficient state. However, improvement of symptoms precedes resolution of MRI findings.

Prognostic Factors

Factors associated with a poor prognosis include

- Heavy alcohol intake (MAST-C >= 6)
- Extra callosal lesions on MRI
- GCS<8, severely disturbed consciousness
- Features of cortical dysfunction

II. Conclusion

MBD is a rare demyelinating neurological condition, conventionally associated with chronic alcoholism, but can also be seen in cases of chronic malnutrition, as was noted in the atypical etiology case. Typical MRI findings include lesions of corpus callosum and subcortical white matter – easily identifiable on MRI. MRI with diffusion weighted imaging can help in early diagnosis and timely intervention. Timely intervention can result in almost complete resolution of MR changes and patient may have a good outcome. Delay in accurate diagnosis may result in coma, death or other serious neurocognitive disturbances. MRI also helps to predict the prognosis of diagnosed cases and rules out other differential diagnosis of MBD.

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