Marchiafava-Bignami Disease (MBD) and Diffusion Tensor Image (DTI) Tractography

Priscilla Chukwueke, Anne Kleiman, Leszek Pisiński

Abstract

Marchiafava-Bignami Disease (MBD) is a rare central nervous system (CNS) disease characterized by demyelination of the corpus callosum. It is mostly found in men with alcohol use disorder and malnutrition with cases reported worldwide across all races. The onset of the disease may be sudden presenting with stupor, coma or seizures while some may present with gait abnormality (spasticity), psychiatric problems, hemiparesis, aphasia, apraxia and incontinence with a resultant high morbidity and mortality rates. Case description: patient is a 30 year old left handed African-American, who presented with c/o altered mental status, urinary incontinence, slurred speech and left-sided weakness. The diagnosis of MBD was confirmed with DTI Tractography which showed significantly diminished commissural fibers extending to the right central semiovale lesion, near absent or significantly diminished commissural fiber extending through the corpus callosum indicating demyelination. Discussion: MBD is often an incidental diagnosis with high morbidity and mortality. This is different from previous cases because of earlier onset as opposed to onset around age 45, rapid recovery and minimal disability as he could walk independently before discharge from hospital. This case also shows added benefit of the DTI tractography in the diagnosis of MBD.

Key words: Marchiafava-Bignami Disease (MBD), demyelination, central nervous system, tractography, semiovale lesion, corpus callosum

Declaration of interest: none

Priscilla Chukwueke, MD, MPH; Anne Kleiman, MD; Leszek Pisiński, MD

Corresponding author
Priscilla Chukwueke
Harlem Hospital Center / Columbia University New York
Email: kaypc2002@yahoo.com

Introduction

Marchiafava- Bignami disease (MBD) is a rare central nervous system (CNS) disease characterized by demyelination of the corpus callosum mostly found in men with alcohol use disorder and malnutrition. The onset of the disease may be sudden presenting with stupor, coma or seizures while some may present with gait abnormality (spasticity) and may also include psychiatric problems, hemiparesis, aphasia, apraxia and incontinence.

This is a debilitating disease and prior to the advent of CT scan and MRI, patients with the disease were found at autopsy. Of the 250 cases reported in 2001, 200 died, 20 had a favorable outcome and 30 were severely disabled. With the advent of CT and MRI, diagnosing this disease is now possible and it has been shown to respond to treatment with intravenous multivitamins B complex, Methyl prednisone, folate and in some cases amantadine. There are two types; A and B. Type A has been shown to have worse prognosis with 81% ending up with long-term disability and 21% mortality while type B has 19% with long-term disability and no mortality. At first this was thought to affect only Italian men (Fauci et al. 2008) but now there has been reports of affected people from all over the world and across all races (Ault 2012). Common diagnostic workup includes CBC, urine toxicology, CT of the brain to rule out bleeding, spinal tap when bleeding has been excluded by CT, EEG to evaluate seizures and MRI which is the most sensitive diagnostic tool.

Objectives

To bring to the awareness of clinicians the possibility of MBD in a patient with alcohol use disorder and malnutrition who presents with neurological disorders.
- The show the importance of DTI in confirming the diagnosis
- The inform the reader about the possibility of an early onset of MBD
- To show the common signs and symptoms of MBD

Case presentation and methods of evaluation

Patient is a 30 year old left handed African-American with history of hypertension, diabetes type I, hypothyroidism, alcohol use disorder, who was brought in by ambulance as a transfer from Columbia University Medical Center (CUMC), New York with c/o altered mental status first reported four days prior to presentation. He was found on the floor by his uncle, was incontinent of urine, had slurred speech and left-sided weakness.
Differential diagnosis

At CUMC, he was seen by neurology and these diagnoses were considered: seizures with post ictal state versus stroke, Cytomegalovirus (CMV), toxoplasmosis, herpes simplex virus (HSV) encephalitis, meningitis, HIV encephalopathy, and JCV. He was empirically started on Ceftriaxone, ampicillin, and acyclovir, as meningitis was being ruled out. Lumbar puncture and CT brain without contrast were done and results were unremarkable. MRI showed bilateral centrum ovale restricting lesion with restriction in splenium of the corpus callosum. Subsequent to the MRI result, MBD was added to the differential and biopsy of the brain was offered, but his mother decided to sign him out from CUMC and transferred him to our facility – Harlem Hospital Center, New York.

On presentation, he had altered mental status, followed a few commands, speech was slurred, left facial asymmetry, left hemiparesis, cerebellar gait, was aggressive, agitated and verbalized suicidal ideation. Neurology was consulted and EEG was ordered which showed abnormal findings consistent with structural abnormality. Lab results was significant for anemia Hb/Hct 12.5/38.4, Albumin was low 2.4 which may be a sign of malnutrition in a young person. HSV PCR was negative, VDRL: negative, CMV & JCV: negative, HIV: negative, Toxoplasmosis negative. He was discharged to home. Psychiatrist and was discharged to home. therapist, occupational therapist, physical therapist, and climb stairs. By day 10, he was cleared by speech therapist, occupational therapist, physical therapist, and psychiatrist and was discharged to home.

Imaging studies

- CT brain w/o contrast: No acute ischemic stroke or hemorrhage
- CT angio: Not diagnostic due motion artifact
- MRA head: No gross aneurysm or vessel wall irregularity
- MRA neck: No stenosis
- MRI W/WO contrast: diffusion abnormality with associated restriction involving splenium symmetrically as well as subcortical white matter of the centrum semiovale right greater than the left side with no associated abnormal enhancement, no mass effect, and differential diagnosis was MBD versus diffuse encephalitis/encephalopathy with reversible splenial lesions versus osmotic demyelinating disease.
- Fluid attenuated inversion recover (FLAIR) showed three lesions:
  - 1. Right centrum semiovale dominant lesion, 2. Left centrum semiovale small lesion and 3. Large lesion in callosal body.
- DTI Tractography: Showed significantly diminished commissural fibers extending to the right central semiovale lesion. Near absent or significantly diminished commissural fibers extending through the splenium of corpus callosum indicating demyelination.
- Fractional Anisotropy (FA) on DTI. Absence of transverse (red-color coded fibers in splenium of corpus callosum).

Discussion

As stated previously, MBD is a rare case and often an incidental diagnosis with high morbidity and mortality. This is different from previous ones because of earlier onset as opposed to onset around age 45, rapid recovery and minimal disability as he could work independently before discharge from hospital. This case also shows added benefit of diffusion tensor image (DTI) tractography in the diagnosis of MBD as clinical presentation was consistent with radiological findings. At first MBD was one of the many differential diagnosis but with DTI it became confirmed. His presentation matches the type A which has high disability rate and mortality, but early diagnosis may have played a role in the prognosis of this patient as he responded to treatment and was commenced early on rehabilitation. Though not the first diagnosis to think about when a patient has alcohol use disorder and presents with loss of consciousness and left-sided paresis, urine incontinence and slurred speech that mimics a stroke, but clinicians and radiologists should keep MBD at the back of their minds when patients with alcohol use disorder and malnutrition presents with the above symptoms.

DTI takes away guess work from the diagnosing.

Why is this a clinical novelty?

- MBD is a rare case and often an incidental diagnosis with high morbidity and mortality.
- This is different from previous ones because of earlier onset (age 30) as opposed to onset around age 45, rapid recovery and minimal disability as he could work independently before discharge from hospital.
- This case also shows added benefit of diffusion tensor image (DTI) tractography in the confirmatory diagnosis of MBD as clinical presentation was consistent with radiological findings. His presentation matches the type A which has high disability and mortality rate, but early diagnosis may have played a role in the prognosis of this patient as he responded to treatment and was commenced early on rehabilitation.
Figure 1. Ax. FLAIR: Hyperintense lesions in bilateral centrum semiovale (Lt > Rt).

Figure 2. Ax. Diffusion tensor imaging (DTI) fractional anisotropy (FA) map: absence/abnormality of red color coded horizontal fibers within splenium of corpus callosum.

Figure 3. Ax. Flair: Hyper intense lesion involving splenium of the corpus callosum.

Figure 4. Ax. T1 contrast enhanced: absence of abnormal enhancement with splenium of corpus callosum.

Figure 5. DTI Tractography: Paucity of commissural fibers in the right centrum semiovale and splenium of corpus callosum.

Figure 6. DTI Tractography: Paucity of fibers in the bilateral corticospinal tracts (Rt > Lt).
References


