Case Report: Creutzfeldt-Jakob Disease

Cameron Manchester M.D.[1], Maximillian Tjauw[2], Iwan Tjauw M.D.[1]
1 Integris Baptist Medical Center, Oklahoma City, Oklahoma
2 University of Pennsylvania, Philadelphia, Pennsylvania

Introduction

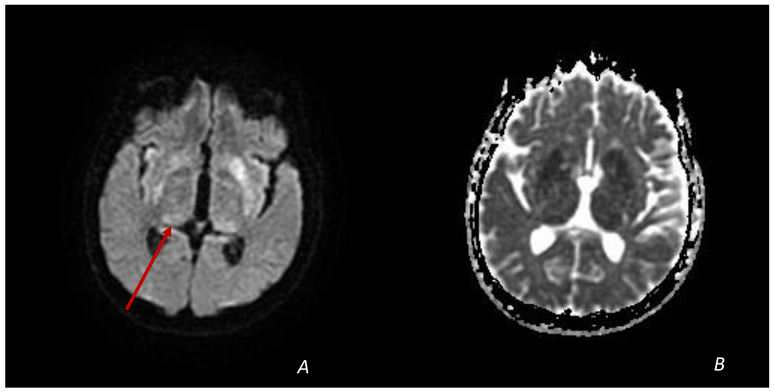
Creutzfeldt-Jakob Disease (CJD) is a rapidly progressive neurodegenerative prion disorder, which is always fatal. CJD incidence rate is around 1 per million individuals each year. Patients affected are typically in the 7th decade, with median survival of 5 months and 90% mortality within the first year.[1] Clinical presentation includes the hallmark rapidly progressive dementia, myoclonus, visual abnormalities, ataxia and behavioral changes with progression to akinetic mutism in late stage.[2] Diagnostic testing includes electroencephalogram (EEG), MRI of the brain, and cerebrospinal fluid analysis.

Case

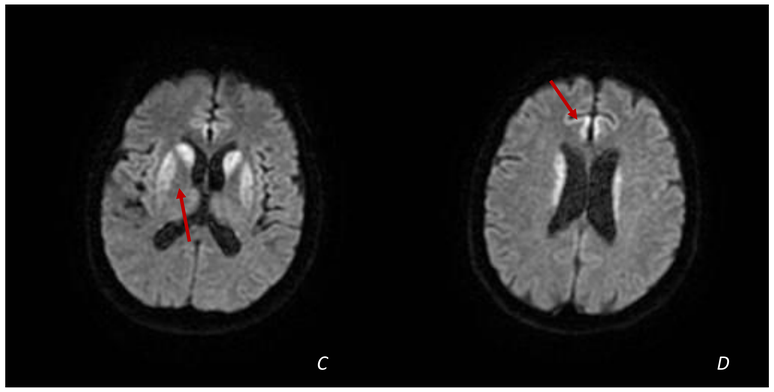
The patient is a 77 year old male with a past medical history of dementia and hypertension presenting with altered mental status. Family history is negative for dementia. He had no prior neurological deficit and no significant travel history, however his family reports declining neurological status over the preceding three months prior to presentation. The patient is unable to give any history. According to the family, he had sudden onset dizziness five months ago for which he was treated for atrial fibrillation. Symptoms progressed rapidly to ataxia, dysarthria, memory loss, cognitive decline, and generalized weakness. The family denies depression and mood disorders. The patient was unable to ambulate at the time of the emergency medical services arrival. Upon physical examination, he was awoken by voice only and showed symptoms of expressive aphasia. He showed increased tone in all four extremities without tremor. His cranial nerve exam was normal. His EEG featured triphasic waves, which can be seen in CJD. A lumbar puncture was not performed.

Imaging

Initial imaging evaluation in the emergency department was performed by a non-contrast CT of the head demonstrating nonspecific white matter changes. Typical CT evaluation in patients with CJD is negative for abnormalities, or CT may have nonspecific atrophy.[3] MRI brain was then performed. Restricted diffusion seen on DWI and ADC affects the bilateral pulvinar nuclei, basal ganglia, and frontal cortices.



MRI brain: *A,* Diffusion Weighted Images (DWI) and *B,* Apparent Diffusion Coefficient (ADC). DWI shows the classic symmetric hockey stick sign (red arrows) of restricted perfusion resulting in increased signal on DWI in the bilateral thalamic pulvinar nuclei. ADC mapping confirms restricted diffusion.



Areas of symmetric restricted diffusion in combination with the thalami include the basal ganglia *(C),* and in this case the frontal cortex *(D).*

Discussion

Creutzfeldt-Jakob disease is part of a group of fatal neurodegenerative disorders caused by proteinaceous infectious particles, or prions, termed transmissible spongiform encephalopathies.[4] Types of CJD include sporadic, which is the cause of 85% of cases, familial, or acquired. The pathophysiology involves formation of the abnormally folded PrPsc protein from the normal PrPc type that then serves as a template for pathologic conformation for other PrPc proteins. These changes cause the classic spongiform changes and neuronal loss.[1] Clinically, patients present with rapidly progressing dementia, and can include other neurological abnormalities including myoclonus, visual changes, cortical blindness, ataxia, and akinetic mutism in late disease.[1,4] Diagnostic testing includes electroencephalography (EEG), cerebral spinal fluid (CSF) analysis, and MRI of the brain. EEG evaluation typically has periodic triphasic sharp wave complex, occurring at a rate of approximately 1/second. EEG findings occur on average 3.7 months after symptom onset and mean survival after EEG abnormalities begin is about 8 weeks.[1] CSF biomarkers for CJD appear as a result of neuronal destruction. The 14—3-3 protein is positive in 92-96% of sporadic CJD cases. The Tau protein is also implicated in CJD with 81% sensitivity.[1] Our patient did not have CSF analysis. MRI findings typically demonstrate the pulvinar sign, which is restricted diffusion within the bilateral pulvinar nuclei of the thalami, in 90% of cases.[5] Restricted diffusion may also be seen in the bilateral basal ganglia and the cortical gray matter. Diffusion weighted imaging is considered superior to other sequences in early disease, but FLAIR hyperintensities in the basal ganglia can also be seen.[1]

Contact

Cameron Manchester M.D.
Integris Baptist Medical Center
Cmanchester24@gmail.com

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