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Creutzfeldt Jakob Disease: Progressive Neurological Decline

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Creutzfeldt-Jakob Disease: Progressive Neurological Decline

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Introduction

- Prion Diseases are neurodegenerative diseases that have prolonged incubation periods, but progress rapidly once clinical symptoms appear. (1)
- 5 Prion diseases currently recognized: Kuru, Creutzfeldt-Jakob disease (CJD), Variant Creutzfeldt-Jakob disease (vCJD) Gerstmann-Straussler-Scheinker syndrome (GSS), and Fatal Familial insomnia (FFI). (1,2)
- Creutzfeld-Jakob Disease is the most common prion disease which can be caused by Sporadic, Familial, Iatrogenic, or Variant forms of CJD.
- We will present a case report about a patient who presented with significant, worsening neurological deficits and was discovered to have Creutzfeldt-Jakob disease.

Background

- Approximately one case of sporadic CJD occurs per 1,000,000 population per year with a worldwide distribution. (1)
- The mean age for the onset of disease is between 57 and 62 years, although rare cases in young adults and those over 80 years of age. (1)
- Rapidly progressive mental deterioration and myoclonus are the two primary clinical manifestations of sCJD.
- Myoclonus, mainly elicited by startle reflex, is present in more than 90 percent of patients at some point during the illness but may be absent at presentation. (3)
- Extrapyramidal signs (ie: hypokinesia and cerebellar manifestations-ie-nystagmus and ataxia) occur in approximately 2/3 of patients and are the presenting symptoms in 20 to 40 percent. (2)
- Due to the time-sensitive nature of CJD, it is imperative to have careful clinical examination and early performance/ interpretation of diagnostic tests, including electroencephalography, quantitative assessment of the surrogate markers 14-3-3, tau, and of the prion protein in the CSF, and neuroimaging. (2)

Case Description

- 58 year old female with a past medical history of essential hypertension, hyperlipidemia, and chronic back pain s/p spinal fusion presented to the Emergency Department with progressive worsening neurologic symptoms since November 2018 had roughly 3 week history of worsening right-sided tremor, slurred speech, difficulty with ambulation, and RLE paresthesias.
- Patient and her husband state patient has had slow progression of memory difficulties, to develop focal right sided weakness and tremors and more recently, slurred speech and right arm dystaxia/incoordination.
- Medical/Surgical History significant for C-spine surgery and fusion in May 2018.
- Neurology was consulted on date of admission for suspicion for underlying demyelinating disease and subsequently Infectious Disease was consulted.
- Thorough workup including CT brain, MRI brain, MRA neck/brain, EEGs, LP, heavy metals, toxicology, other infectious/viral labs ordered.
- While inpatient, patient's symptoms worsened with increasing hemiballismus, startle myoclonus, and worsening memory recall.

Results

- MRI Brain obtained revealed bilateral supratentorial hyperintense diffuse signal abnormalities noted involving basal ganglia and cortical ribboning noted in L > R hemispheres. (Figure 1)
- First EEG showed no abnormalities, however, second EEG revealed spike and wave complexes which are highly associated with prion disease.
- Clinical symptoms including neurological deficits with rapid deterioration in conjunction with imaging-placed CJD as most probable diagnosis.
- On Day 15, LP CSF fluid resulted from National Prion Laboratory for 14:3:3 and returned positive with >98% sensitivity for CJD.

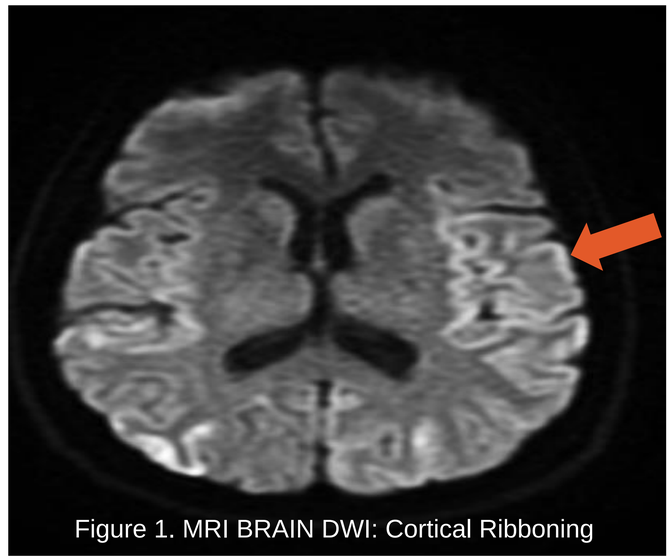

Figure 1. MRI BRAIN DWI: Cortical Ribboning

Conclusion

- With thorough history gathering, assess the mode of patient's CJD-sporadic, recent corneal transplant, neurosurgical intervention, etc.
- CJD should be highly suspected in a patient with symptoms of both rapidly progressive dementia and myoclonus.
- Once a CJD diagnosis has high probability, it is important to have prognostication discussions with patient and family in order to educate the patient on progression of disease and provide resources.
- Physicians and laboratories are required to report to local city/state County Health Department every prion disease case, most within 24-48 hours of identification or diagnosis.
- There is no effective treatment for CJD which is uniformly fatal. Death usually occurs within one year of symptom onset with a median disease duration of six months.

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