“All in the Family”: Diagnosis of Familial Creutzfeldt-Jakob Disease, A Case Report

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Case Diagnosis

Creutzfeldt-Jakob disease (CJD), familial variant.

Vignette

65-year-old male with 18-year history of polyneuropathy presents with 2-months of acutely progressive ataxia in all extremities. Physical exam showed muscle atrophy and fasciculations, lower extremity weakness with hyporeflexia, and length-dependent sensory loss. Empiric treatment with methylprednisolone significantly improved his ataxia. He was discharged to Shirley Ryan AbilityLab where he functionally improved through steroid-taper and was discharged home with a caregiver. One month later, he was readmitted with acute worsening of cognitive impairment and lower extremity strength. Plasma exchange (PLEX) provided no symptomatic improvement. Notably, the patient’s mother and sister died with death certificates listing cause of death as CJD.

Laboratory

Cerebral spinal fluid sent to The National Prion Disease Pathology Surveillance Center:

- T-tau 7685 pg/mL (ref 0-1149 pg/mL)
- 14-3-3 Positive (ref negative)

Interpretation: positive for T-tau and 14-3-3 protein with likelihood of prion disease >98%.

Studies

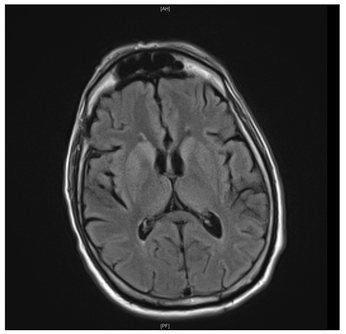

Figure 1: MRI Brain T2-weighted fluid-attenuated inversion recovery (FLAIR) image showing increased diffusion restriction with hyperintense signal involving the bilateral caudate nuclei and lentiform nuclei (mostly putamen). Not pictured: similar changes in the bilateral hippocampi. These findings are not typical of neurofascin neuropathic process but consistent with prion disease.

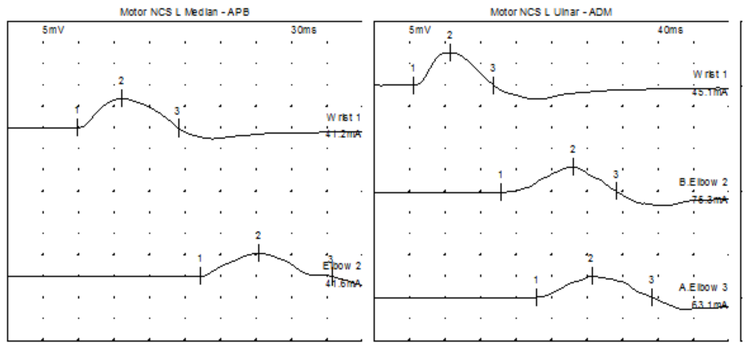

Table 1: Electromyography and nerve conduction studies (NCS) performed on the left upper and lower extremity finding diffusely slowed motor conduction velocities that are relatively uniform at 20 m/s. Shown here are the NCS for the left median and left ulnar nerves. Findings are concerning for severe demyelinating polyneuropathy.

Learning points

Epidemiology: CJD is a transmissible, progressive neurogenerative disorder, characterized as a transmissible spongiform encephalopathy. Subcategories include:

- sporadic (85-95% of cases)
- genetic (5-15%)
- acquired (<1%).[1]

Familial CJD (fCJD) occurs when CJD is inherited (genetic).

Presentation: memory loss, ataxia, tremors, and weakness. However, dementia in fCJD progresses more slowly and presents later in life when compared with sporadic CJD.

In this case of fCJD, the patient presented with an insidious polyneuropathy and did not develop rapidly progressive dementia until years later. In reported cases of CJD, patients have presented similarly and with EMG studies that resemble chronic inflammatory demyelinating polyneuropathy.[2,3] These presentations are supported in transgenic mice models overexpressing wild-type prion protein.[4]

Treatment:

- In this case, steroids alleviated symptoms.
- PLEX had little functional benefit.
- Rehabilitation should focus on self-care and mobility deficits.

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References

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