A Rare Presentation of Creutzfeldt-Jakob Disease Mimicking an Inflammatory Demyelinating Polyneuropathy

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Introduction

- Creutzfeldt-Jakob disease (CJD) is the most common of the human prion diseases[1]
- CJD is part of a group of neurodegenerative disorders caused by the cerebral deposition of a misfolded, β-sheet enriched, and partially protease-resistant isoform of the cellular prion protein[1]
- Common clinical symptoms of CJD include neuropsychiatric symptoms, myoclonus, nystagmus, and ataxia[1]
- Recent development and validation of the prion real-time quaking-induced conversion (RT-QuIC) assay, the first specific in vivo diagnostic test for CJD, has significantly increased the potential for an early accurate clinical diagnosis[1]

Case Description

- 65-year-old male with a history of an essential tremor was admitted to an acute care hospital with progressive lower extremity weakness and ataxia
- Nerve conduction study confirmed the presence of a demyelinating polyneuropathy, with conduction blocks, temporal dispersion, prolonged terminal latencies, and reduced conduction velocities
- Laboratory testing was positive for anti-neurofascin antibody
- The patient was treated with intravenous immunoglobulin and intravenous corticosteroids with mild improvement in ataxia
- He was discharged to acute inpatient rehabilitation where he demonstrated limited functional improvements and required re-admission to acute care for interval decline with new cognitive and memory impairments
- MRI brain revealed changes concerning for CJD and further family history revealed multiple family members who have died of a familial form of CJD
- Cerebrospinal fluid studies were positive for RT-QuIC, elevated T-tau protein, and positive 14-3-3 protein. Likelihood of prion disease was identified as > 98%.

Discussion

- Neurofascin antibody has been associated with chronic inflammatory demyelinating polyneuropathy[2]
- Neurofascin antibody is associated with axon loss, sensory ataxia, weakness and occasional demyelinating features as were observed with this patient[2]
- Peripheral nervous system involvement has been documented with a diagnosis of CJD[1,3], although less common and should prompt additional assessment to rule-out an alternative diagnosis

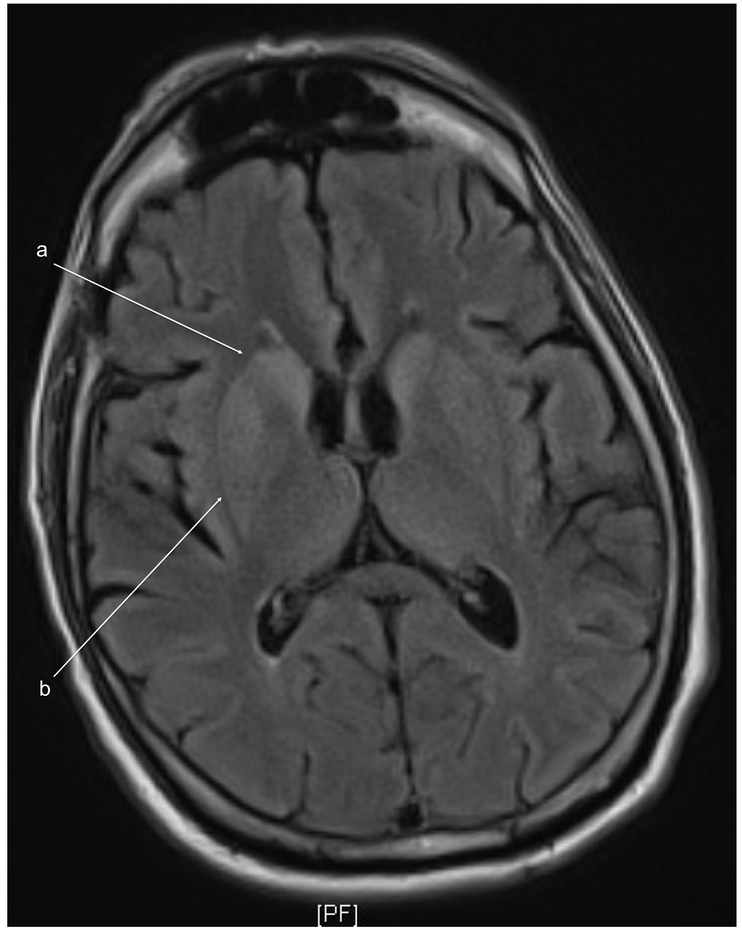

Figure 1

There is increased diffusion restriction with faint T2/FLAIR hyperintense signal involving the bilateral caudate nuclei (a) and lentiform nuclei mostly the putamen (b), new from two months prior

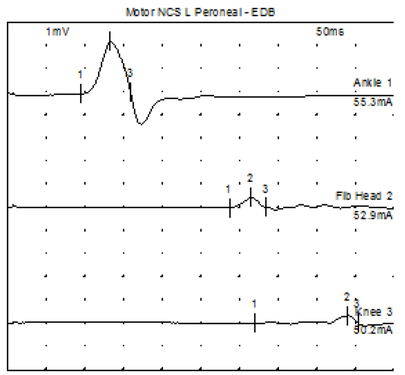

Figure 2

Focal conduction block of the peroneal-extensor digitorum brevis motor nerve in the foreleg accompanied by temporal dispersion and prolonged distal latency

Nerve / Sites	Latency (ms)	Reference (ms)	Amplitude (mV)	Reference (mV)	Duration (ms)	Distance (mm)	Velocity (m/s)	Reference (m/s)
L Median - APB								
Wrist	5.89	≤4.00	6.8	≥5.0	8.54	50		
Elbow	16.30		5.5		11.09	280	26.9	≥49.0
L Ulnar - ADM								
Wrist	4.48	≤3.10	7.7	≥7.0	9.01	50		
B.Elbow	14.27		6.0		13.02	220	22.5	≥49.0
A.Elbow	18.28		5.0		13.02	110	27.4	
L Peroneal - EDB								
Ankle	9.48	≤6.00	2.3	≥2.5	6.41	80		
Fib Head	28.70		0.5		4.69	420	21.9	≥39.0
Knee	31.98		0.3		13.33	95	29.0	
L Peroneal - Tib Ant								
Fib Head	5.21	≤4.50	2.1	≥3.0	8.33			
Knee	8.65		2.0		6.30	95	27.6	
L Tibial - AH								
Ankle	NR	≤6.00	NR	≥4.0	NR	80		

Table 1

Motor Nerve Conduction Study demonstrating conduction blocks, prolonged terminal latencies, and reduced conduction velocities

Conclusion

- This case highlights an extremely rare clinical presentation of Creutzfeldt-Jakob Disease, mimicking an inflammatory demyelinating polyneuropathy with positive anti-neurofascin antibody
- Although rare, it is important to understand this association to ensure proper evaluation and diagnosis in a timely fashion

References

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