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Case Report

Acute Onset of Creutzfeldt-Jakob Disease Resembling Stroke in Absence of Diagnostic EEG Findings

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Abstract

We report a case of Creutzfeldt-Jakob disease with acute onset of symptoms with EEG negative for PSWC, resembling a cerebrovascular accident. The patient was diagnosed with sporadic CJD by positive RT-QUIC, positive 14-3-3 protein, T-Tau protein of 14560 pg/mL, brain magnetic resonance imaging (MRI) demonstrating restricted diffusion on DWI in bilateral frontal, temporal, occipital cortex, caudate and putaminal nuclei, as well as T2 FLAIR hyperintensities in the bilateral cerebral cortex and basal ganglia. The entire course of the disease from onset to death was 52 days.

***Keywords:** CJD, Acute onset, EEG, Myoclonus, Ataxia, Caudate nucleus, Ribboning*

Introduction

Creutzfeldt-Jakob disease (CJD), also known as subacute spongiform encephalopathy, is a rare neurodegenerative human prion disease caused by a rapid misfolding of proteins resulting in neuronal loss, proliferation of astrocytes, and extra-neuronal vacuolization which leads to the "spongiform" appearance of the cortex.[1] The disease is always fatal with rapidly progressive dementia of subacute onset. EEG findings are significant for generalized Periodic Sharp Wave Complexes (PSWC).[1]

We report a case of a patient who presented with an acute (vs. sub-acute) onset of symptoms with EEG negative for PSWC, resembling a cerebrovascular accident. Patient was diagnosed with sporadic CJD by positive RT-QUIC (96% sensitivity, 100% specificity), positive 14-3-3 protein (88% sensitivity, 96% specificity), T-Tau protein of 14560 pg/mL, brain magnetic resonance imaging (MRI) demonstrating restricted diffusion on DWI in bilateral frontal, temporal, occipital cortex, caudate and putaminal nuclei, as well as T2 FLAIR hyperintensities in the bilateral cerebral cortex and basal ganglia.[2,3]

Case Presentation

A 55-year-old Caucasian male with no medical history presented with acute onset of declining mental acuity, sensory deficits, memory loss, bradyphrenia, severe ataxia, unilateral myoclonus, and suicidal ideations. Patient's symptoms began suddenly without previous symptomatology. The acute onset of symptoms was triggered by an episode of what the patient described as heat exhaustion. He complained of suddenly feeling mental fogginess, generalized weakness with right facial numbness, which he believed was due to COVID-19 infection, but subsequent viral testing was negative. The patient had been a strict vegetarian for the past 18 years, had not traveled outside of the country, had not participated in any unusual or risky behavior and described himself as an introvert. Our patient was a highly intelligent man with an IQ of 145, who worked as a computer programmer.

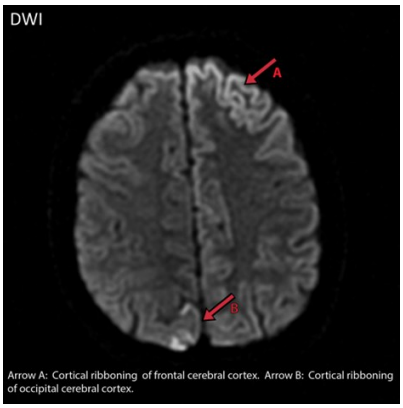
The patient stated that approximately two days after the initial onset of symptoms, he developed faint twitching in his right arm and leg, difficulties with thinking, composing emails, expressing himself as well as difficulties with coordination and gait. He was unable to walk independently, or prepare meals, which resulted in a 12 lb. weight loss. After approximately 10 days of progressively rapid mental and physical decline, he sought medical attention at an Emergency Department, where head CT ruled out stroke or any acute abnormalities, and subsequently patient was discharged the same day. The patient did not agree with the emergency physician's lack of concern or diagnosis, and followed up with an outpatient neurologist who referred him to our hospital for direct admission. Upon admission and during the following two weeks of hospitalization, he remained alert and oriented with normal reading and verbal comprehension with debilitating ataxia, worsening speech latency, thought blocking, concentration, and memory, as well as right-sided sensory deficits and more pronounced myoclonus. He developed profound anxiety and repeatedly stated he was "going to die". He also demonstrated and verbalized suicidal ideations. Inpatient psychiatry service was consulted and the patient was placed twice on suicide watch under Baker Act 52 (Florida Mental Health Act). During his third and last week of hospitalization, myoclonus became severe in all extremities and the patient developed akinetic mutism.

On initial physical examination patient was alert and oriented with short and long term memory impairment, bradyphrenia, word finding difficulties, facilitatory paratonia, and cross response inhibition. Cranial nerves were intact except for right-sided CN V1-3 sensory loss. Other neurologic deficits included subjective diminished sensation of right arm and leg to light touch, vibration and pin prick, poor right finger to nose coordination, and severe ataxia. Motor strength was MRC 5/5 in all muscle groups and normal reflexes.

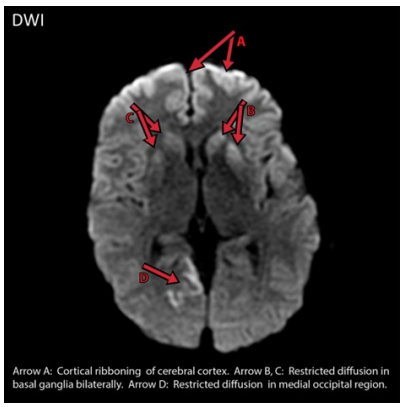
Diagnostic Tests

MRI of the brain revealed bilateral occipital cortical atrophy, ribboning-like areas of restricted diffusion on DWI within the bilateral cerebral cortex, frontal and occipital lobes bilaterally, and left caudate and putaminal nuclei. T2 FLAIR hyperintensities were evident within the same areas, particularly within the left basal ganglia as well as left dorsomedial thalamic nucleus.

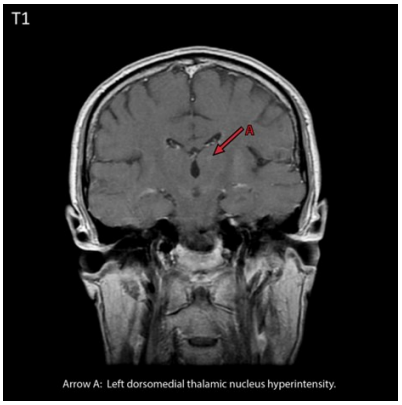
Additionally, a standardized Electroencephalogram (EEG) was performed. Activity was symmetrical with normal amplitude 7Hz PDR. Bi-frontal leads recorded larger amplitude theta and delta wave slowing. Generalized Periodic Sharp Wave Complexes (PSWC) were not recorded, and no epileptiform discharges were detected. Finally, cerebral spinal fluid (CSF) tested positive for RT-QUIC, 14-3-3 protein positive, and T-TAU protein of 14,560 PG/mL. (Range 0- 1149).



Arrow A: Cortical ribboning of frontal cerebral cortex. Arrow B: Cortical ribboning of occipital cerebral cortex.



Arrow A: Cortical ribboning of cerebral cortex. Arrow B, C: Restricted diffusion in basal ganglia bilaterally. Arrow D: Restricted diffusion in medial occipital region.



Arrow A: Left dorsomedial thalamic nucleus hyperintensity.

Outcome

Patient met the CDC diagnostic criteria for Sporadic Creutzfeldt-Jakob disease, and was ultimately discharged to Hospice. Two days after discharge from our hospital the patient died. The entire course of the disease from the first onset of symptoms to death was 52 days.

Discussion

CJD is the most common prion disease and is characterized by sub-acute onset with clinical features of non-specific early symptoms followed by rapid cognitive decline, as well as cerebellar ataxia, visual disturbances, and myoclonus.[1] The initial presentation of this case was unusual due to the sudden onset of executive functioning deficit, ataxia, and sensory deficits which initially mimicked a cerebrovascular accident. However, brain MRI and the increasing myoclonus suggested differential diagnosis of Wilson's disease, heavy metal poisoning, and CJD. A similar case of another highly intelligent patient who presented with an acute onset of neurobehavioral disorder mimicking stroke like symptoms triggered by general anesthesia was described by Hohler A.D. et al.[4]

Diagnostic criteria for CJD can be met by using EEG, neuroimaging, CSF analysis, and clinical symptoms, but one should not disregard the possibility of CJD if EEG is negative for the highly specific PSWC[1,5]. EEG is a useful diagnostic tool, however, it can be non-specific, and therefore, a high index of suspicion is needed in cases where clinically CJD is suspected.[5] Bortone et al studied 15 CJD cases during 1975-1991 with confirmed diagnosis in 12 patients on post-mortem biopsy.[5] Out of the 15 cases, PSWC were recorded in 14 patients and one patient's EEG did not record PSWC until the end stage of the disease.[5] Furthermore, studies documented by Appleby, A.S., et al suggested the Brownell -Oppenheimer variant of sporadic CJD also lacks the highly specific PSWC, however, this phenotype lacks basal ganglia hyperintensities on MRI imaging, has the longest 92 day average survival time with the longest survival time of 421 days, and presents only with ataxia, therefore, we cannot suggest our patient suffered from the Brownell-Oppenheimer variant.[6]

The only definite diagnosis for CJD is brain biopsy, however, since the sample is not always obtained from the affected part of the brain, and CJD brain tissue is highly infectious, biopsies and autopsies are discouraged. In our case, patient's symptoms of ataxia, myoclonus, rapid cognitive decline, neuroimaging significant for CJD and positive CSF studies met the diagnostic criteria for sporadic CJD despite the absence of PSWC on EEG.

Conflict of Interest

None

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