MEDDOCS Open Access Publisher

ISSN: 2637-885X

Journal of Radiology and Medical Imaging

Open Access | Case Report

Creutzfeldt-Jakob disease (CJD): A Case Report and the Role of Radiological Imaging

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Received: Oct 25, 2024

Accepted: Nov 11, 2024

Published Online: Nov 18, 2024

Journal: Journal of Radiology and Medical Imaging

Publisher: MedDocs Publishers LLC

Online edition: http://meddocsonline.org/

Abstract

Creutzfeldt-Jakob Disease (CJD) is a rare and fast-progressing neurodegenerative disorder caused by abnormal prion proteins. It can present with symptoms, such as psychiatric or neurological condition. This case report describes a middle-aged woman with Creutzfeldt-Jakob Disease (CJD), initially thought to have psychiatric disorder due to symptoms of agitation and cognitive decline. Her condition rapidly worsened, and MRI revealed brain abnormalities characteristic of CJD. Despite the lack of Cerebrospinal Fluid (CSF) analysis and typical EEG findings, a diagnosis of sporadic CJD was made through clinical and radiological evaluation. The case emphasizes the importance of looking for clinical findings and MRI to support the diagnosis CJD when CSF analysis is not possible. This case also highlights the need for early recognition of symptoms and the role multidisciplinary approach when managing patients with this fatal disease.

Introduction

CJD appears to be caused by an abnormal infectious protein called a prion. These prions accumulate at high levels in the brain and cause irreversible damage to nerve cells. It is characterized by rapidly progressive dementia, motor deficits, and often psychiatric manifestations. Diagnosing CJD can be challenging, as initial symptoms may mimic other neurological or psychiatric conditions.

Case presentation

A middle-aged female presented to the emergency department with a two-week history of progressive agitation, and cognitive decline. Her past medical history was significant for hypothyroidism and diabetes, with no prior psychiatric or neurological disorders. Initial neurological examination was unremarkable with no signs of meningitis and no focal neurological deficits were observed.

Routine blood tests showed an elevated white cell count (WCC 26.3), with a lymphocyte count of 15.8. Blood cultures showed no growth and PCR test for viral infections were negative. All other blood tests were unremarkable. A CT scan of the head was performed and showed no acute abnormalities. MRI was done and it showed signs associated with meningoencephalitis. Additional blood tests for autoimmune encephalitis screen were also unremarkable and patient was started on Acyclovir. A However despite completing the anti-viral course, the patient had no clinical improvement.

Based on the patient's behavioral changes and collateral history of recent job loss and stress, the psychiatric team cognitive assessment using the Mini-Addenbrooke's Cognitive Examination revealed significant impairment, with a score of 8/30. As a result, she was diagnosed with acute stress disorder or dissociative symptoms. She was started on lorazepam and olanzapine for management of agitation.

Cite this article: Ali A, Myo H, Vijayaragavan P, Sheikh S. Creutzfeldt-Jakob Disease (CJD): A case report and the role of radiological imaging. J Radiol Med Imaging. 2024; 7(2): 1098.

Progression of the disease

Over the subsequent months, the patient's condition deteriorated with worsening confusion, agitation and aggressive behavior. She also developed a shuffling, ataxic gait. Neurological examination at this stage raised concern for an underlying neurodegenerative or inflammatory process. An MRI of the brain was ordered, and EEG was also requested, CSF analysis could not be performed due to the patient's congenital spina bifida and tethered spinal cord, complicating the lumbar puncture procedure.

MRI Report

The initial MRI scan showed bilateral cerebral gyral restricted diffusion, initially interpreted as suggestive of meningoencephalitis. However, following a detailed review and with further clinical correlations, the MRI findings were revised to demonstrate multifocal bilateral cerebral cortical diffusion restriction, with involvement of the frontal lobes, parieto-occipital regions, and the right posterior temporal lobe. Further abnormalities were also identified in the corpus striatum, predominantly on the right side, and in the pulvinar regions of the thalami, radiological features suggestive of CJD. Despite the absence of Periodic Lateralized Epileptiform Discharges (PLEDs) on EEG, which are often associated with CJD, the constellation of MRI findings and the patient's rapidly neurological exam findings and progressive neurological decline led to a diagnosis of CJD, made on the balance of probability after extensive discussions and input from the CJD team.

Given the absence of familial history or exposure risks, the patient was most likely suffering from the sporadic form of the CJD (sCJD).

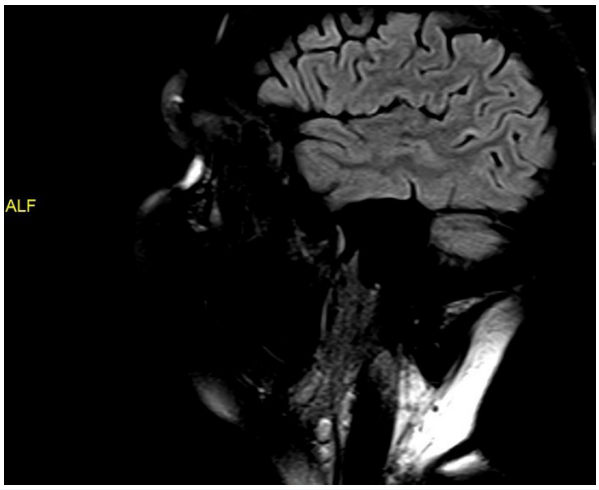

Figure 3: FLAIR: Subtle gyral hyperintensity.

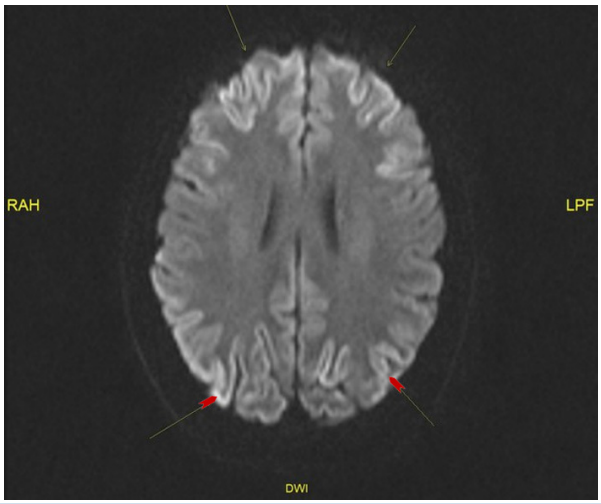

Figure 1: DWI image: Matched Gyral restricted diffusion with ADC.

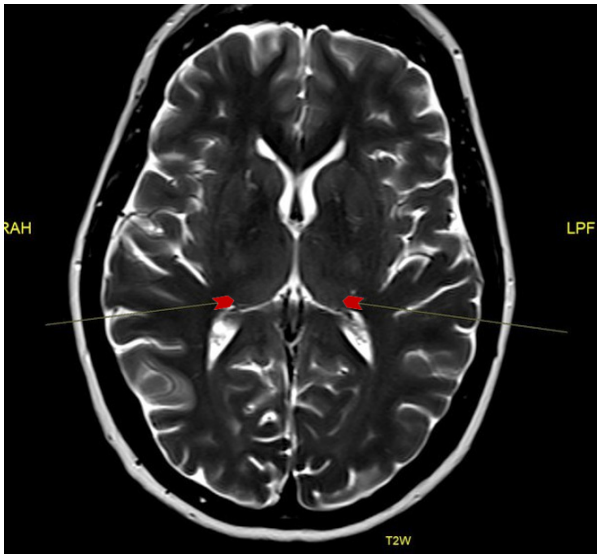

Figure 4: T2W image: Bilateral pulvinar area abnormality showing subtle hyperintensity.

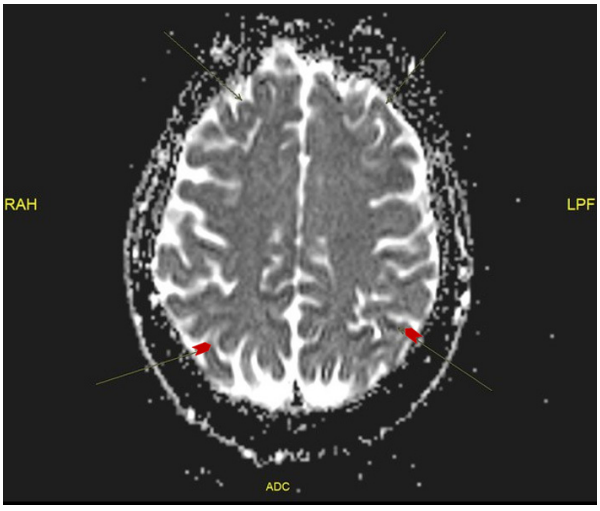

Figure 2: ADC image: Matched Gyral restricted diffusion with DWI.

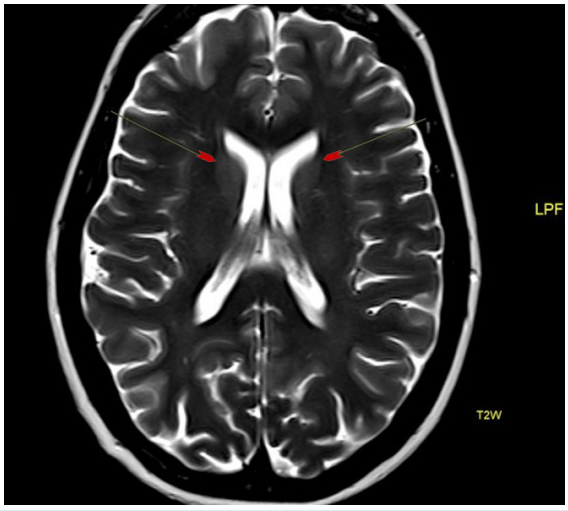

Figure 5: T2W image: Corpus striatum changes more on the right compared to the left.

Management and Outcome

Following the diagnosis of the sporadic form CJD, the patient's neurological status continued to deteriorate. She had a reduction in her mobility, increasing dependence on others for all activities of daily living, and finally becoming bedbound. After multidisciplinary discussions, including with the neurology, palliative care, and CJD specialist teams, the patient was discharged to a nursing care facility with a focus on palliative care management.

Discussion

CJD remains a diagnostic challenge due to its variable clinical presentation and the often-non-specific nature of early symptoms. This case highlights the importance of considering CJD in patients with rapidly progressive neurological and cognitive deterioration, particularly when initial diagnoses, such as psychiatric disorders, do not fully explain the progression of symptoms.

Conclusion

This case report highlights the complexity of diagnosing CJD, particularly when patients initially present with psychiatric symptoms. In the absence of confirmatory tests such as CSF analysis, neuroimaging can be crucial in guiding diagnosis. Early recognition is critical for the timely initiation of appropriate care, including palliative measures, given the aggressive and invariably fatal nature of CJD. This case underscores the importance of considering prion diseases in the differential diagnosis of rapidly progressive cognitive and neurological decline.

Differential diagnosis and investigations

Bacterial/Viral Encephalitis - Blood cultures showed no growth, VZV, HSV-1 and HSV-2 DNA PCR, Meningococcal and pneumococcal PCR - all negative. In addition, unresponsive to antiviral medication.

Autoimmune Encephalitis - NMDA RECEPTOR ABS, CONTACTIN2 ASSCOCIATED AB (anti-Caspr2 antibodies), LEUCINE RICH GLIOMA I1 AB (anti-Lgi1 antibodies), GLUTAMATE RECEPTOR ABS (anti-AMPA1 antibodies, anti-AMPA2 antibodies), anti-GABA antibodies, ANTI MOG ANTIBODIES -all negative.

Hashimoto encephalopathy - autoantibodies negative and unresponsive to steroid.

Learning Points

Atypical Presentations of Creutzfeldt-Jakob Disease (CJD)

CJD can initially present with psychiatric symptoms, such as agitation, aggressive behavior, and cognitive decline mimicking conditions like acute stress disorder or dissociation. This emphasizes the need to consider neurodegenerative causes in cases of rapidly progressive psychiatric or cognitive symptoms.

Diagnostic Challenges in CJD and role of MRI

The absence of definitive EEG findings, such as periodic lateralized epileptiform discharges (PLEDs), and the inability to perform cerebrospinal fluid (CSF) analysis, highlight the challenges in diagnosing CJD in this case. MRI is critical in identifying characteristic features of CJD, such as multifocal cortical diffusion restriction, particularly involving the frontal, parieto-occipital, and temporal lobes, along with involvement of the thalamus and corpus striatum, pointed to a diagnosis of CJD.

The Need for Multidisciplinary Collaboration Accurate diagnosis and management of CJD require input from various specialties, including neurology, radiology, psychiatry, and palliative care. Early involvement of a multidisciplinary team can help in making timely diagnoses and initiating appropriate care, especially in rapidly progressive and fatal diseases like CJD.

Palliative Care in CJD

Given the rapid neurological deterioration seen in CJD, early palliative care involvement is essential for managing the patient's quality of life and supporting the family. This case highlights the importance of transitioning to palliative care when curative options are not available, ensuring the patient receives compassionate and supportive care.

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