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CASE REPORT

A Rare Case of Creutzfeldt-Jakob Disease Presenting with Amyotrophic Lateral Sclerosis

Yethindra Vityala[1,*], Tugolbai Tagaev[1], Altynai Zhumabekova[1], Manjula Shantaram[2]

[1] *Honorary International Faculty, AJ Institute of Medical Sciences and Research Centre, Mangalore, Karnataka, India*
[2] *AJ Research Centre, AJ Institute of Medical Sciences and Research Centre, Mangalore, Karnataka, India*

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* *Corresponding author.*
Yethindra Vityala
yethindravityala10@gmail.com

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ABSTRACT

Creutzfeldt-Jakob disease (CJD) is a rare, fatal neurodegenerative condition with rapid dementia and motor issues. A 76-year-old woman developed progressive quadriparesis over three years, beginning in the lower limbs and extending to the upper limbs. Clinical assessment revealed upper and lower motor neuron involvement, including spasticity, hyperreflexia, bilateral Babinski's sign, and fasciculations. Brain magnetic resonance imaging revealed bilateral thalamic lesions, indicating possible transmissible spongiform encephalopathy. Neuropsychological testing revealed fronto-subcortical syndrome with mild cognitive impairment. Neuropathology confirmed CJD with subcortical and motor cortex involvement, early-stage amyotrophic lateral sclerosis, argyrophilic grain disease (stage III), and moderate Alzheimer's changes (A2B1C2). Cognitive symptoms were primarily attributed to the CJD. This case demonstrates the diagnostic challenges of atypical CJD presentations mimicking amyotrophic lateral sclerosis, highlighting the importance of neuroimaging, clinical, and neuropathological correlation in elderly patients with motor and cognitive decline.

Keywords: Creutzfeldt-Jakob disease; Amyotrophic lateral sclerosis; Magnetic resonance imaging; Neuropathology

CASE REPORT

A 76-year-old woman presented with a history of quadriparesis from three years ago, initially in the lower extremities and ascended towards the upper extremities (symmetrical and progressive). Neurological examination revealed mild quadriparesis, spasticity, hyperreflexia, bilateral Babinski sign, and fasciculations in the infraspinatus muscle. Magnetic resonance imaging revealed bilateral thalamic lesions, where a broad differential diagnosis was made, which included transmissible spongiform encephalopathies (TSEs). Because of this atypical finding, magnetic resonance imaging could identify it as a variant of Creutzfeldt-Jakob disease (CJD).

Neuropsychological studies revealed fronto-subcortical syndrome characterized by mild cognitive impairment (MCI) with memory loss and abnormalities in visuospatial capacity, and fronto-executive function. The neuropathological study revealed TSEs with predominantly subcortical and motor cortex involvement, which is congruent with CJD. Furthermore, it was associated and congruent with amyotrophic lateral sclerosis (ALS), incipient stage III argyrophilic grain disease per staging of argyrophilic grains[1], and Alzheimer's disease neuropathologic change, classified as stage A2B1C2 (Braak stage I, Thal phase 3, Consortium to Establish a Registry for Alzheimer's Disease score moderate) of the National Institute on Aging and Alzheimer's Association classification system (2012)[2].

The presence of amyotrophy in CJD as a first symptom is very rare[3]. The patient's symptoms initially resembled a clinical presentation of ALS. Moreover, prion transmission originated in the anterior horn cells, ascending to the corticospinal tracts, and eventually to the brain. The presence of MCI clearly attributed to CJD. Although changes in Alzheimer's disease were evidenced in neuropathology, these were insufficient to describe the cognitive symptoms.

DISCLOSURES

Conflicts of interest

The authors declare that there are no conflicts of interest.

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