A rare case of Creutzfeldt-Jakob disease presenting with amyotrophic lateral sclerosis

Yethindra Vityala[1], Elmira Mamytova[2], Dzhamalbek Turgumbaev[3], Altynai Zhumabekova[4], Tugolbai Tagaev[2], and Sagynali Mamatov[2]

[1]International Higher School of Medicine, International University of Kyrgyzstan
[2]I K Akhunbaev Kyrgyz State Medical Academy
[3]S.B. Daniyarov Kyrgyz State Medical Institute for Retraining and Advanced Studies
[4]City Maternity Hospital No. 2

March 31, 2021

Abstract

Here, we present 76-year-old woman, who presented with a history of quadriparesis from three years ago, initially in the lower extremities and ascended towards the upper extremities (symmetrical and progressive). Magnetic resonance imaging revealed bilateral thalamic lesions (Figure 1), which could identify it as a variant of Creutzfeldt-Jakob disease.

A rare case of Creutzfeldt-Jakob disease presenting with amyotrophic lateral sclerosis

Yethindra Vityala[1*], Elmira Mamytova[2], Dzhamalbek Turgumbaev[3], Altynai Zhumabekova[4], Tugolbai Tagaev[5], Sagynali Mamatov[6]

[1]Department of Pathology, International Higher School of Medicine, International University of Kyrgyzstan, Bishkek, Kyrgyzstan

[2]A.N. Murzaliev Department of Neurology and Clinical Genetics, I.K. Akhunbaev Kyrgyz State Medical Academy, Bishkek, Kyrgyzstan

[3]Department of Neurology, Neurosurgery and Psychiatry, S.B. Daniyarov Kyrgyz State Medical Institute for Retraining and Advanced Studies, Bishkek, Kyrgyzstan

[4]Obstetrician-gynecologist, City Maternity Hospital No. 2, Bishkek, Kyrgyzstan

[5]Department of Public Health and Healthcare, I.K. Akhunbaev Kyrgyz State Medical Academy, Bishkek, Kyrgyzstan

[6]Department of Hospital Internal Medicine, Occupational Pathology with a Course of Hematology, I.K. Akhunbaev Kyrgyz State Medical Academy, Bishkek, Kyrgyzstan

Running title: Magnetic resonance imaging in diagnosis of Creutzfeldt-Jakob disease

***Corresponding author:** Vityala Yethindra, Department of Pathology, International Higher School of Medicine, International University of Kyrgyzstan, Bishkek, Kyrgyzstan, Email: yethindravityala10@gmail.com, Contact: +91 9121925658

ABSTRACT

Here, we present 76-year-old woman, who presented with a history of quadriparesis from three years ago, initially in the lower extremities and ascended towards the upper extremities (symmetrical and progressive).

Magnetic resonance imaging revealed bilateral thalamic lesions (Figure 1), which could identify it as a variant of Creutzfeldt-Jakob disease.

Keywords: Neurology, Creutzfeldt-Jakob disease, Amyotrophic lateral sclerosis, Magnetic resonance imaging

Key clinical message

In this study, magnetic resonance imaging helped a broad differential diagnosis, which included Creutzfeldt–Jakob disease (CJD) and the presence of argyrophilic grain disease contributed to mild cognitive impairment which further attributed to CJD.

CASE SUMMARY

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 (Figure 1), 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.

ACKNOWLEDGMENTS

The authors acknowledge their patient for kind cooperation and for providing consent.

FINANCIAL SUPPORT

No financial support was received for the study.

CONFLICT OF INTEREST

The authors declare no conflicts of interest.

AUTHOR CONTRIBUTIONS

Conception, design of the work, manuscript preparation, and data acquisition: Yethindra Vityala, Elmira Mamytova, Dzhamalbek Turgumbaev, Altynai Zhumabekova, Tugolbai Tagaev, Sagynali Mamatov, Clinical management: Yethindra Vityala, Elmira Mamytova, Dzhamalbek Turgumbaev, Altynai Zhumabekova, and Manuscript preparation and data acquisition: Yethindra Vityala, Tugolbai Tagaev, Sagynali Mamatov.

DATA AVAILABILITY STATEMENT

Data are available from the corresponding author upon reasonable request.

REFERENCES

1. Saito Y, Ruberu NN, Sawabe M, et al. Staging of argyrophilic grains: an age-associated tauopathy. *J Neuropathol Exp Neurol.* 2004; 63: 911-8.
2. Hyman BT, Phelps CH, Beach TG, et al. National Institute on Aging-Alzheimer's Association guidelines for the neuropathologic assessment of Alzheimer's disease. *Alzheimers Dement.* 2012; 8: 1-13.
3. Panegyres PK, Armari E, Shelly R. A patient with Creutzfeldt-Jakob disease presenting with amyotrophy: a case report. *J Med Case Rep.* 2013; 7: 218.