

Creutzfeldt-Jacob Disease: a Case Report

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Abstract- Creutzfeldt-Jacob Disease is a prion disease which has a wide range of clinical presentations. Its diagnosis is not simple and clinical manifestation along with EEG, MR imaging findings and cerebrospinal fluid (CSF) analysis should be considered for a definite diagnosis. A-50-year-old woman referred with cognitive impairment, myoclonic jerks, mutism and difficulty in swallowing to our clinic. EEG (Electroencephalography) results showed bilaterally periodic sharp and slow-wave discharges. Protein 14-3-3 in CSF was detected. Magnetic resonance imaging (MRI) findings revealed hyperintensity of the caudate and putamen in diffusion-weighted imaging (DWI), T2 Weighted (T2W) sequences and Fluid-attenuated inversion-recovery (FLAIR) images. Patients who have progressive dementia should be evaluated by means of MR imaging and CSF analysis for CJD specific proteins should be considered.

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Introduction

Creutzfeldt-Jacob disease (CJD) is a rare, neuro-degenerative, prion disease with an incidence of one per million individuals per year (1). It occurs mostly as sporadic form (90%) followed by familial (15%) and acquired forms (5%) (2). Progressive dementia, visual disturbances, myoclonus, extrapyramidal and pyramidal signs, and behavioral disturbances are among the most common clinical findings (2). The triad of dementia, ataxia and myoclonus consist the most common clinical symptoms of CJD.

The diagnosis of CJD is a highly challenging issue as, clinical manifestation should be considered along with electroencephalogram (EEG), CSF and serum analysis, neuro-imaging, and prion protein gene and the isoform detection for the definite diagnosis.

Alzheimer disease, Infectious or granulomatous disorders, tumors, Hashimoto thyroiditis, paraneoplastic syndromes, and vasculitis could mimic clinical picture of Creutzfeldt-Jacob disease. Here, we report a CJD case presented with unusual symptoms.

Case Report

The patient was a 50-year-old woman presented to

our clinic with tinnitus, vertigo and ataxia since six months ago. Chiari malformation had been considered in another clinical visit, and she had been a candidate for surgery. During the time before surgery, she got cognitive impairment and developed myoclonic jerks, speech impairment and difficulty in swallowing.

When she attended to our clinic, she was unable to talk. Physical examination showed a conscious, not-oriented female with flat affect who did not obey the rules.

Vital signs were all normal; mydriatic reactive pupils, symmetric nasolabial folds and rigidity in all extremities detected. Deep tendon reflex exam showed hyperreflexia (3+), and she presented abnormal Babinski reflex in both sides. Cerebellar, sensory and gait examinations were impossible due to patient's non-cooperation. EEG (Electroencephalography) results showed bilaterally periodic sharp and slow-wave discharges.

Magnetic resonance imaging (MRI) findings revealed hyperintensity of the caudate and putamen in diffusion-weighted imaging (DWI), T2 Weighted (T2W) sequences and Fluid-attenuated inversion-recovery (FLAIR) images.

Cerebrospinal fluid (CSF) examination revealed no pleocytosis but showed positive 14-3-3 protein by

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Western blot method. Based on clinical manifestations and para-clinic findings, she was diagnosed with sporadic CJD and died within nine months after her first complaints.

Discussion

CJD is a rare, fatal neuro-degenerative disease with distinctive clinical and pathological features (1).

Sporadic CJD usually appears at 50–70 years of age consisting 90% of all cases with CJD. The diagnosis of Sporadic CJD is based on clinical symptoms along with laboratory tests including an electroencephalogram (EEG), CSF and serum analysis for specific proteins such as S-100 protein, Tau protein, 14-3-3 protein, imaging and pathology (1).

Behavioral disturbances, progressive dementia, visual disturbances, myoclonus, cerebellar, akinetic mutism, pyramidal, and extrapyramidal symptoms are among clinical findings while one third of patients present fatigue, sleep disturbances and decreased appetite. Dementia, ataxia, and myoclonus are considered as CJD triad. Ataxia is a late-stage symptom of the disease, and myoclonus has been suggested as a reaction to auditory and tactile stimuli (3).

On T2W MR imaging, hyperintense areas could be found in the caudate nucleus, putamen, globus pallidus and thalamus. Diffusion-weighted MRI may indicate bilateral symmetrical hyperintense signals in the basal ganglia and/or cortex cerebri. DW MRI has been considered as a proper modality for CJD diagnosis in the early phase of the disease (4) Shiga et al., reported sensitivity and specificity of DW MRI in diagnosing CJD as 92.3% and 93%, respectively (4). Cerebral atrophy can be another MRI finding.

14-3-3 protein is a protein which is released from damaged neurons into CSF and is a good marker for CJD with the sensitivity and specificity reported near

94% and 84%, respectively (5). It could be detected in other diseases such as viral encephalitis, Hashimoto encephalitis and amyotrophic lateral sclerosis. Therefore, it has to be considered along with other clinical and para-clinical findings for CJD diagnosis.

EEG is a diagnostic modality with sensitivity near 64% and specificity between 74 and 91% (5). In our case, EEG showed typical changes for CJD.

Gozke et al., reported a Seventy year-old man attended to their clinic with behavioral changes, insomnia, irritability and weakness. EEG findings were non-specific, and DW MRI showed hyperintense signal changes in the cortex. Protein 14-3-3 was detected (3).

Our case showed typical EEG and MRI signs and positive 14-3-3 protein, which are important clues for CJD diagnosis.

Conclusion: Patients who have progressive dementia should be evaluated by means of MR imaging and CSF analysis for CJD specific proteins should be considered.

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