Case Report

# Sporadic Creutzfeldt Jakob Disease: A Patient with Dementia and Involuntary Movement Myoclonus

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## Abstract

Creutzfeldt Jakob Disease (CJD) is an incurable, invariably fatal, rapidly progressive neurodegenerative disease caused by an abnormal isoform of a cellular glycoprotein known as the prion protein. CJD occurs worldwide, estimated annual incidence is about one case per million populations per year. Sporadic (sCJD) is a human prion disease; infection with this disease usually leads to death within one year of onset of illness. The characteristic clinical and diagnostic features of rapidly progressive dementia, myoclonus, visual or cerebellar signs, pyramidal and extrapyramidal signs, akinetic mutism and positive result on the presence of 14-3-3 protein in CSF assay, typical EEG features and MRI findings of brain are highly suggestive of diagnosis. Biopsy of brain for histopathological examination is more specific and confirmatory for diagnosis. This article reports a 65 years old lady of sCJD who was diagnosed by characteristic findings of MRI of brain, Electro-encephalography (EEG) and cerebrospinal fluid (CSF) assay at National University Hospital (NUH) Singapore and now admitted at United Hospital Limited (UHL) Dhaka for palliative and supportive management.

***Key-words:*** Creutzfeldt Jakob Disease (CJD), Human prion disease, Neurodegenerative disease.

## Introduction

Epidemiologically sCJD is the most common human prion disease, rare in worldwide at a rate of one case per million populations per year, most frequently in patients 55-65 years of age. It can occur in people younger than 55 years of age but are extremely rare[1]. In more than 85 % of cases, the duration of CJD is less than one year after the onset of symptoms.

Types of CJD include Sporadic (sCJD) caused by a mutation arising in an individual of unknown reason and this accounts for 85% cases of CJD[2]. Varient (vCJD) caused by consuming food contaminated with prion. Familial (fCJD) caused by an inherited mutation and accounts for the other 15% cases of CJD. Iatrogenic caused by contamination of tissue from infected person usually as a result of medical procedure e.g. corneal and meningeal transplant, blood transfusion. Diagnosis solely based on typical clinical manifestation, characteristic EEG findings, presence of 14-3-3 protein in CSF assay[3], MRI of brain-high signal intensity in caudate nucleus and putamen symmetrically on T2WI and DWI are most sensitive[4]. The present manuscript reports encounter in National University Hospital, Singapore and United Hospital Limited, Dhaka known as sporadic CJD.

## Case Report

A 67-year-old highly educated active lady admitted at neurology department of UHL Dhaka with 02 months history of rapidly progressing personality change, became dementic with loss of memory, impairment of judgment and intellectual functions, became anxious, depressed and aphasic with inappropriate sound. She also had visual disturbances with hallucination, become ataxic with incoordination of her gait. For the last one and half months she developed myoclonus that persists during sleep, provoked by loud sound and bright light but no history of seizure attack. Clinically GCS>8, pupil bilaterally equal and reacting to light, speech-aphasic with inappropriate sound and diminished cough reflex. No cranial nerve dysfunction with normal fundus, motor function normal with ataxic gait. Sensory function could not be able to assess. All the relevant investigations blood CBC, RBS, creatinine, and lipid profile, liver, renal and thyroid function were normal. Serum ANA, Anti-dsDNA antibody, TSH, anti-TPO antibody, TSH receptor antibody, serum autoimmune encephalitis panel including PET, CT, FDG whole body scan were within normal limit. The presence of 14-3-3 protein in CSF analysis, typical EEG findings with periodic generalized sharp wave's complex, MRI of brain revealed symmetrical high signal intensity in DWI and T2WI in caudate and lentiform nucleus and left fronto-tempero-parietal cortex. She had been thoroughly evaluated in NUH Singapore and concluded the diagnosis of Sporadic CJD.

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## Discussion

Sporadic CJD is a human prion disease rapidly progressive, invariably fatal, neurodegenerative disease that occurs worldwide. The majority of CJD patients usually die within one year of onset of illness. CJD is classified as a transmissible spongiform encephalopathy along with other prion disease that occur in human and animals[5].

Patient initially became symptomatic with rapidly progressive dementia leading to memory loss, personality change, impairment of judgment and intellectual power. Other feature of anxiety, depression, paranoia, and psychosis. This is accompanied by physical problem such as speech impairment, myoclonus, ataxia with changes in gait, rigid posture and seizure attack. At last the patient died with complications of pneumonia and respiratory failure. There is no test to confirm the diagnosis of CJD, only a brain biopsy or an examination of brain tissue after death (autopsy) can confirm the presence of Creutzfeldt-Jakob disease. The following investigations can help to diagnose such as EEG which shows characteristic generalized periodic sharp waves pattern (Fig-1), CSF assay for the presence of 14-3-3 protein[6], MRI of brain revealed symmetrical high signal intensity in caudate nucleus and putamen on DWI and T2WI (Fig-2) and Immunohistochemical analysis of brain tissue shows the marked accumulation of protease resistance prion protein. Brain biopsy is the definite diagnostic test which shows classic appearance of spongiform changes in gray matter, presence of rounded vacuoles which appear glassy or eosinophilic (Fig-3). Neuronal loss and gliosis are seen[7].

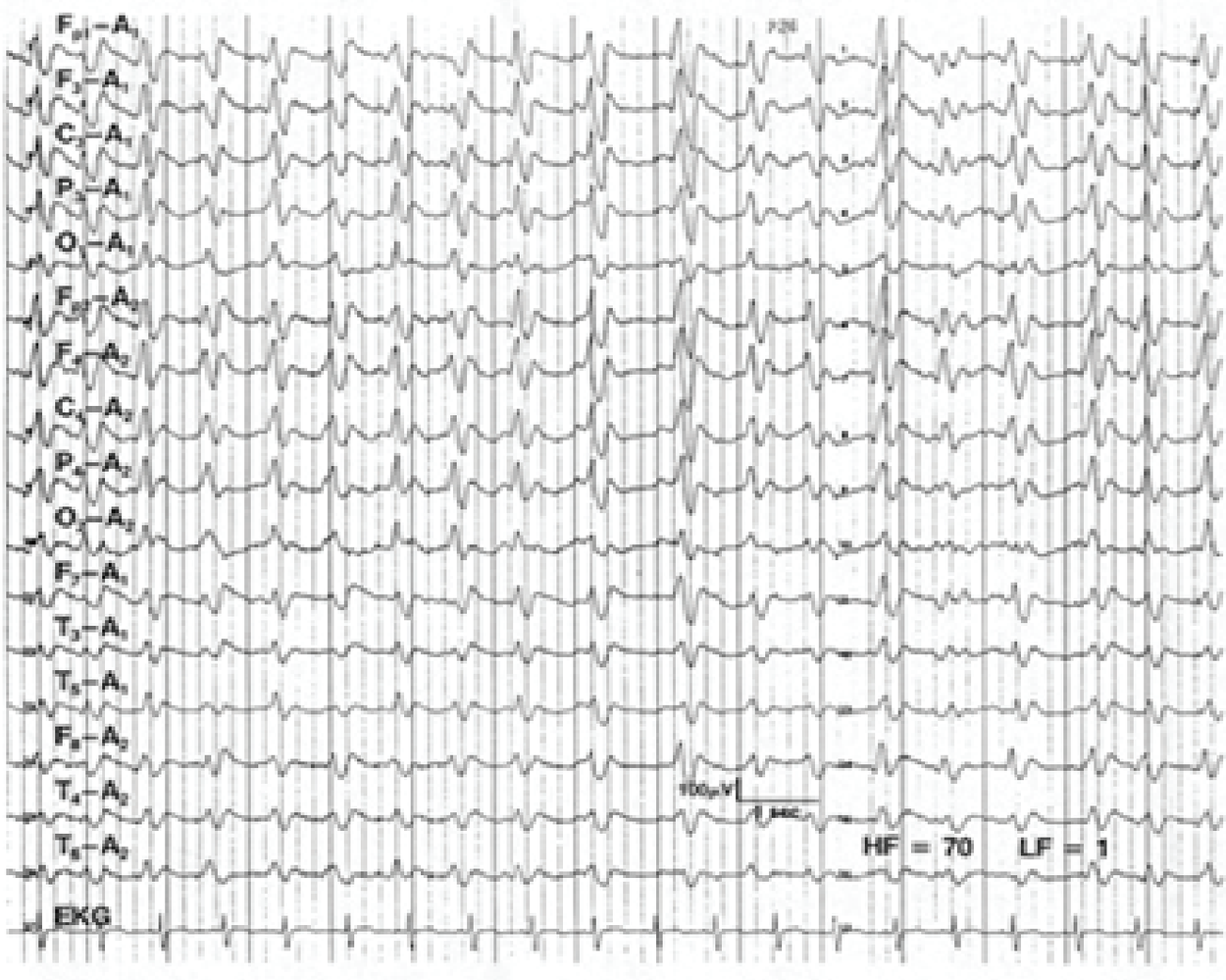


**Fig-1:** EEG showing characteristic generalized periodic sharp waves pattern

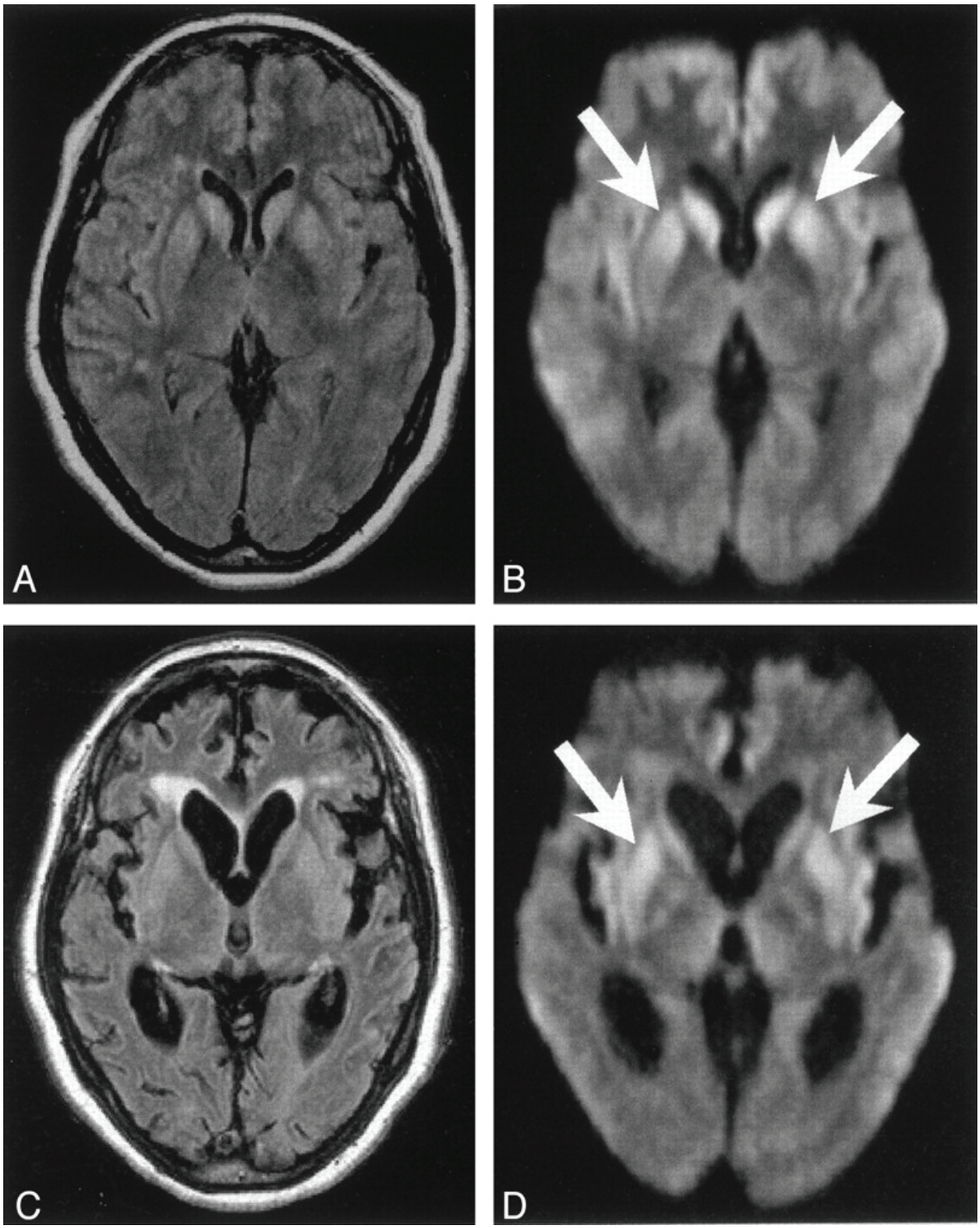


**Fig-2:** MRI of brain revealed symmetrical high signal intensity in caudate nucleus and putamen on DWI and T2WI

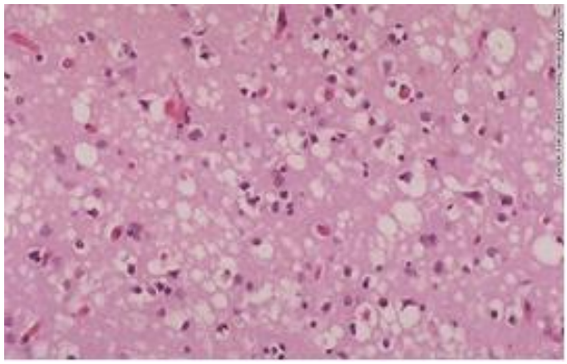

**Fig-3:** Immunohistochemical analysis of brain tissue shows the marked accumulation of protease resistance prion protein. Brain biopsy is the definite diagnostic test which shows classic appearance of spongiform changes in gray matter, presence of rounded vacuoles which appear glassy or eosinophilic. Neuronal loss and gliosis are seen.

There is no cure of CJD. No drug yet is available to stop the progression of disease. However some medications are in clinical trial. Pentosan polysulphate may slow the progression of disease. Amphotericin-B and Doxorubicin as yet there is no strong evidence that either drug is effective in stopping the disease. Quinacrine permanently cleared abnormal prion protein from cell culture but had no measurable effect on clinical course of CJD[8]. Aztemizole have anti-prion activity may be effective for the treatment of CJD. Current treatment aims to alleviate symptoms and makes the patient as comfortable as possible.

## Conclusion

Sporadic CJD though is rare but may be missed due to lack of suspicion and diagnostic facilities. Any rapidly progressing dementic patients with myoclonus have to be evaluated with CSF assay, EEG and MRI of brain though no specific treatments are yet available. However extensive research may be effective to find out the curable drugs for CJD management in future.

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