Dementia and Neurocognitive Disorders
2010; 9: 100-2

■ CASE REPORT ■

Focal Dystonia as an Initial Manifestation of Sporadic Creutzfeldt-Jakob Disease with Uncommon MRI Findings

Jihoon Kim, M.D.*, Kee Ook Lee, M.D.*†

Department of Neurology*, Konyang University College of Medicine, Daejeon; Department of Neurology†, Yonsei University College of Medicine, Seoul, Korea

Received : April 19, 2010
Revision received : July 26, 2010
Accepted : August 6, 2010

Address for correspondence

Kee Ook Lee, M.D.
Department of Neurology, Konyang University Hospital, 685 Gasuwon-dong, Seo-gu, Daejeon 302-718, Korea
Tel: +82-42-600-8814
Fax: +82-42-545-0050
E-mail: niceiatros@naver.com

Sporadic Creutzfeldt-Jakob disease (CJD) is a rare progressive spongiform encephalopathy. Involuntary movements commonly occur in the course of sporadic CJD, but focal dystonia alone as an initial manifestation is rare. We described a 40-yr-old man who presented with a 2 week history of focal dystonia in the neck and left arm. Interestingly, diffusion-weighted images revealed high signal intensities in the right temporal, posterior parietal, and occipital cortices rather than in the striatum.

Key Words: *Creutzfeldt-Jakob disease, Dystonia, Magnetic resonance image*

Sporadic Creutzfeldt-Jakob disease (CJD) is a rare progressive spongiform encephalopathy, which is characterized by a clinical presentation of rapidly progressive dementia, ataxia, myoclonus, visual disturbances, and various movement disorders.[1] One third of the patients initially present with nonspecific complaints of general weakness, sleep disturbance, or decreased appetite. Another third initially have cognitive deficits, such as memory loss, confusion, or behavioral abnormalities. Remaining patients initially have focal neurologic signs, such as ataxia, aphasia, visual disturbance, or hemiparesis.[2] Although involuntary movements commonly occur in the course of the disease, it is rare for them to occur as an initial manifestation in sporadic CJD.[3]

We report a unique patient who showed focal dystonia as the only presenting symptom of probable CJD and without striatal involvement on brain magnetic resonance imaging (MRI).

CASE REPORT

A 40-yr-old man was admitted to the hospital due to a 2-week history of blurred vision and twisted posture on the neck and left upper extremity. He had no medical illnesses or family history of neurological diseases. He did not have behavioral change or emotional instability. The Mini Mental State Examination (MMSE) was 29 out of 30, which suggested that cognition was normal. His dystonic posture was shown on the left upper extremity; the arm was flexed and abducted, and the wrist was flexed and the ulnar deviated; the fingers and thumb were flexed at rest. He could actively extend his fingers and wrist. Other movement disorders were not shown, including myoclonus and choreoathetosis. On ophthalmological test, there were no obvious abnormalities. Laboratory test results, including complete blood count, erythrocyte sedimentation rate, C-reactive protein, chem-

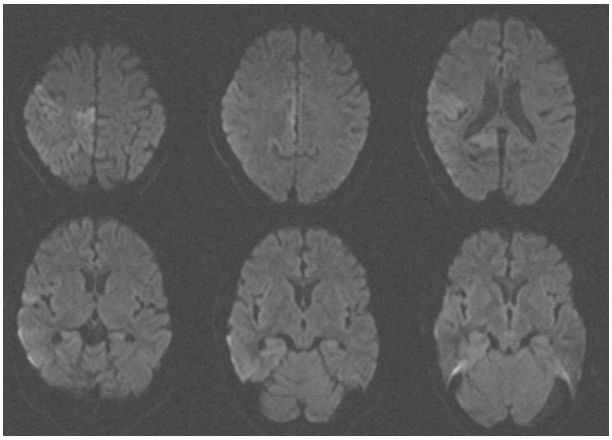

Fig. 1. Initial MRI. Diffusion-weighted images show high signal intensities in the sensory motor cortex, without signal changes in the striatum.

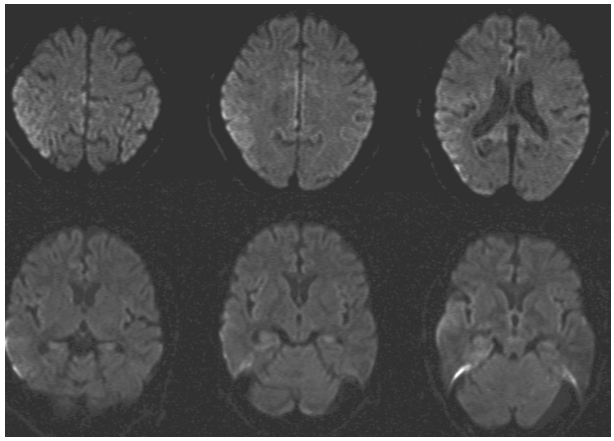

Fig. 2. Follow up MRI, performed on 20th hospital day. Despite focal dystonia is contiLnued, striatal signal changes are not shown on diffusion-weighted images.

istry, vitamin B_{12}, folate, anti-HIV, VDRL, thyroid function test, were normal. A cerebrospinal fluid (CSF) study revealed an opening pressure of 130 mm of CSF, white blood cell (WBC) count 1 cell/mm^3, proteins 85 mg/dL, and glucose 136 mg/dL. The CSF studies including herpes simplex virus, cytomegalovirus, Epstein-Barr virus polymerase chain reactions, cryptococcal antigen, cytology, acid-fast bacilli, fungal, viral, and bacterial stains and cultures were unremarkable. The brain MRI revealed high signal intensity lesions in the right temporal, posterior parietal and occipital lobe without striatal involvement on diffusion-weighted images (Fig. 1). An electroencephalogram (EEG) showed periodic lateralized epileptiform discharges in the right hemisphere with background slowing. The western blot test for the 14-3-3 protein, in the CSF, was positive, while the prion protein gene (PRNP) mutation was negative. During the next 3 weeks, progressive behavioral change and cognitive decline developed. Follow-up MRIs, performed on the 9th and the 20th hospital day, also showed the absence of striatal involvement (Fig. 2), despite the continuation of focal dystonia. Three months later, his cognitive impairment and dystonia progressively worsened.

DISCUSSION

Approximately 90% of the patients present movement symptoms during the course of CJD.[1,4] The most common movement disturbances during the disease course were myoclonus (80%), gait disturbance (80%), and cerebellar ataxia (77%). Dystonia occurs in only 20% of the patients who present with movement symptoms.[5]

Although the frequency of movement symptoms in CJD increases with disease duration, only a very small portion of patients had movement symptoms as an initial manifestation.[3] In a previous epidemiological study, only 5% of the patients presented with involuntary movements at the initial stage.[6]

Furthermore, focal dystonia is rarely presented in the early stages of the disease.[7-9] In previously reported cases, dystonia was usually associated with other movement disorders, such as focal myoclonus, choreoathetoid movement, tremors, or bradykinesia.[8,10,11] In this case, the patient presented with an isolated focal dystonia, and other movement disorders were not shown.

In sporadic CJD, a high signal in the striatum, the cerebral cortex, and, to a lesser extent, in the thalamus are the typical findings of a brain MRI.[12] The frequent striatal involvement and its dysfunction are considered to be key factors in the pathogenesis of involuntary movements.[1] In previous similar cases of CJD, presented with focal dystonia, the MRI findings usually showed an increased signal in the striatum.[7-9] In our case, the patient presented with focal dystonia as an initial manifestation and it continued several

weeks, but striatal signal change has not been shown on the initial or follow-up MRI. It is conceivable that dystonia is related with abnormal activity of the contralateral sensory motor cortex (SMC). The impairment of GABAergic intracortical inhibition has been proposed as a cardinal pathophysiologic feature of dystonia.[13] But, we could not completely rule out the possibility of functional changes in the striatum. To assist in determining the functional correlates of these structural abnormalities, more functional imaging should be used to measure regional cerebral blood flows and dopaminergic function in this case.

REFERENCES

1. Maltête D, Guyant-Maréchal L, Mihout B, Hannequin D. *Movement disorders and Creutzfeldt-Jakob disease: a review. Parkinsonism Relat Disord 2006; 12: 65-71.*
2. Johnson RT, Gibbs CJ Jr. *Creutzfeldt-Jakob disease and related transmissible spongiform encephalopathies. N Engl J Med 1998; 339: 1994-2004.*
3. Brown P, Cathala F, Castaigne P, Gajdusek DC. *Creutzfeldt-Jakob disease: clinical analysis of a consecutive series of 230 neuropathologically verified cases. Ann Neurol 1986; 20: 597-602.*
4. Will RG, Matthews WB. *A retrospective study of Creutzfeldt-Jakob disease in England and Wales 1970-79. I: Clinical features. J Neurol Neurosurg Psychiatry 1984; 47: 134-40.*
5. Edler J, Mollenhauer B, Heinemann U, Varges D, Werner C, Zerr I, *et al. Movement disturbances in the differential diagnosis of Creutzfeldt-Jakob disease. Mov Disord 2009; 24: 350-6.*
6. Will RG, Matthews WB, Smith PG, Hudson C. *A retrospective study of Creutzfeldt-Jakob disease in England and Wales 1970-1979. II: Epidemiology. J Neurol Neurosurg Psychiatry 1986; 49: 749-55.*
7. Lee SH, Suh SI, Koh SB. *Spreading dystonia in probable Creutzfeldt-Jakob disease with serial diffusion-weighted magnetic resonance images. Eur Neurol 2007; 58: 122-4.*
8. Hellmann MA, Melamed E. *Focal dystonia as the presenting sign in Creutzfeldt-Jakob disease. Mov Disord 2002; 17: 1097-8.*
9. Maltête D, Guyant-Maréchal L, Gérardin E, Laquerrière A, Martinaud O, Mihout B, *et al. Hemidystonia as initial manifestation of sporadic Creutzfeldt-Jakob disease. Eur J Neurol 2006; 13: 667-8.*
10. Sethi KD, Hess DC. *Creutzfeldt-Jakob's disease presenting with ataxia and a movement disorder. Mov Disord 1991; 6: 157-62.*
11. Zochodne DW, Young GB, McLachlan RS, Gilbert JJ, Vinters HV, Kaufmann JC. *Creutzfeldt-Jakob disease without periodic sharp wave complexes: a clinical, electroencephalographic, and pathologic study. Neurology 1988; 38: 1056-60.*
12. Tschampa HJ, Zerr I, Urbach H. *Radiological assessment of Creutzfeldt-Jakob disease. Eur Radiol 2007; 17: 1200-11.*
13. Ridding MC, Sheean G, Rothwell JC, Inzelberg R, Kujirai T. *Changes in the balance between motor cortical excitement and inhibition in focal, task specific dystonia. J Neurol Neurosurg Psychiatry 1995; 59: 493-8.*