Sporadic Creutzfeldt-Jakob Disease Presenting with Fixed Focal Dystonia

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ABSTRACT

OBJECTIVE - A wide spectrum of neurologic symptoms occurs during the course of prion disease. We describe a unique case of dystonia being the presenting feature in sporadic Creutzfeldt-Jakob disease.

BACKGROUND - Sporadic Creutzfeldt-Jakob disease is a rare neurodegenerative disease, with many clinical presentations. Rapidly progressive dementia is a main clinical symptom, and movement disorders, specifically myoclonus and gait disturbance, typically occur later in disease.

DESIGN/METHODS - A case from the Movement Disorders Clinic at the Lahey Hospital & Medical Center is reviewed, with clinical presentation, imaging studies, electroencephalogram, laboratory results, and clinical course reviewed.

RESULTS - A 64 year old woman presented to the Movement Disorder Clinic with subacute, progressive, focal dystonia of the right hand. She first noticed diminished sensation in the 4th and 5th digits eight weeks prior to presentation, and later developed cramping and involuntary flexion of the fingers and her hand. There was a high amplitude jerky, irregular action tremor. An EMG of the upper extremity was performed on two separate occasions, and was found to be normal. A brain MRI was unremarkable. A brain SPECT scan showed moderate asymmetric decreased perfusion to the right globus pallidus, and small focal perfusion defects involving the left frontal and parieto-occipital cortices. EOG showed moderate encephalopathy with excessive bifrontal theta and delta activity. Broad range chemistries results were normal, but protein 14-3-3 later returned positive. The patient’s mental status rapidly deteriorated over two months, and she discharged home with hospice care. She died three months later, less than seven months from onset of symptoms.

CONCLUSIONS - Movement disorders are unusual presenting symptoms of CJD. Of these, fixed dystonia is rare, with one case previously reported. In the majority of cases, dystonia is associated with other movement disorders, such as focal jerky myoclonus, choreothetoid movements, postural tremor, alien limb, or bradykinesia.

INTRODUCTION

Sporadic Creutzfeldt-Jakob disease is a rare neurodegenerative disease that presents with a rapidly progressive dementia as an initial clinical symptom. This is combined with a findings of a mixed pattern of the four additional symptoms of myoclonus, visual or cerebellar disturbances, pyramidal or other extrapyramidal dysfunction, and akinetic mutism. Symptoms, however may be variable and include a broad spectrum of movement disorders.

Movement Disorders and focal sensory complaints are rarely the presenting complaint in CJD, although the majority of patients develop myoclonus and extrapyramidal as the disease progresses.

We describe a patient who presented with a fixed focal hand dystonia accompanied by features of a complex regional pain syndrome (CRPS) in the right upper extremity. Her subsequent clinical course and ancillary studies were consistent with probable CJD. This case raises questions about the mechanism by which dystonia and CRPS may coincide, and it highlights a distinctly unusual presentation of prion disease.

CASE

A 64 year-old woman, who as a professional writer, presented to movement disorder clinic with a subacute, progressive, focal dystonia of the right hand. Two months prior, she noticed diminished sensation in the fourth and fifth digits, followed by cramping and involuntary flexion of the fingers, to the point that she could no longer open her hand with other extremities uninvolved. Her medical history was significant for osteoporosis. At the time of presentation, she took no medications and her family history included a brother with spasmodyic dystonia.

Physical examination: Gross inspection revealed that the right hand was markedly swollen, with asymmetric decreased perfusion to the right globus pallidus, and small focal perfusion defects involving the left high frontal and parieto-occipital cortices. EOG showed moderate encephalopathy with excessive bifrontal theta and delta activity. Broad range chemistries results were normal, but protein 14-3-3 later returned positive. The patient’s mental status rapidly deteriorated over two months, and she discharged home with hospice care. She died three months later, less than seven months from onset of symptoms.

Figure 1: SPECT Scan images demonstrate moderate asymmetric decreased perfution to the right globus pallidus as compared to the left. There are also small focal perfusion defects involving the left high frontal, and left parietal, parieto-occipital cortices.

Figure 2: There is development of restricted diffusion within bilateral striatum, caudate-lenticular bridges, left parietal cortex involvement of the left > right cingulate cortex.

DISCUSSION

Movement disorders as a presenting symptom for CJD are rare, as only 1 in 230 describes the symptom onset. The most common movement disorders in CJD patients at presentation include myoclonus (80%), gait disturbances (80%), and cerebellar ataxias (77%). Focal dystonia had previously been reported in cases of sporadic or familial CJD, as well as for the variant CJD. Types of dystonia described include blepharospasm, focal hand dystonia, truncal dystonia and neck dystonia.

In this case, the SPECT abnormalities correlated well with the EEG findings, and preceded the development of characteristic diffusion-weighted MRI changes by several weeks.

REFERENCES