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Rapidly progressive tetraplegia and cognitive deterioration during rehabilitation: A case of neurodegenerative disease

Rehabilitasyon sırasında hızlı ilerleyen tetrapleji ve bilişsel bozulma: Bir nörodejeneratif hastalık olgusu

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Abstract
Human prion diseases are fatal, progressive neurodegenerative disorders caused by neurolytic pathogen proteins, called prions. The most common human prion disease is sporadic Creutzfeldt-Jakob disease, with an approximate annual prevalence of 0.5-1 per million. The symptoms and signs include rapidly progressive dementia, ataxia, myoclonic seizures, akinetic mutism and other neurological and neurobehavioral disorders. The clinical spectrum of Creutzfeldt-Jakob disease is highly variable; therefore it can be difficult to diagnose premortem. This article describes a 78-year-old woman who initially presented with difficulty walking and balance disorder. As a result of the evaluation, the patient was transferred to rehabilitation clinic, with a diagnosis of cervical spinal stenosis. During hospitalization, she showed progressive decline in gait and balance and deteriorated rapidly. The patient was considered to be probable sporadic Creutzfeldt-Jakob disease after further investigations.
Keywords: Neurodegenerative disease, Creutzfeldt-Jakob disease, Rehabilitation

Öz
İnsan prion hastalıkları, prionlar olarak adlandırılan nörolitik patojen proteinlerin neden olduğu ilerleyici nörodejeneratif hastalıklardır. En yaygın insan prion hastalığı sporadik Creutzfeldt-Jakob hastalığı olup, yıllık prevalansı yaklaşık milyonda 0.5-1'dir. Semptomlar ve bulgular; hızla ilerleyen demans, ataksi, miyoklonik nöbetler, akinetik mutizm ve diğer nörolojik ve nörodavranışsal bozuklukları içerir. Creutzfeldt-Jakob hastalığının klinik spektrumu oldukça değişkendir, bu nedenle premortem teşhis etmek zor olabilir. Bu makalede, başlangıçta yürüme zorluğu ve denge bozukluğu ile başvuran 78 yaşında bir kadın hasta anlatılmaktadır. Yapılan değerlendirme sonucunda hasta servikal spinal stenoz tanısı ile rehabilitasyon kliniğine transfer edildi. Hastanede yatışı sırasında, yürüme ve dengesi progresif olarak kötüye giderek bozuldu. İleri tetkiklerden sonra hastanın olası sporadik Creutzfeldt-Jakob hastalığı olduğu düşünülmüştür.
Anahtar kelimeler: Nörodejeneratif hastalık, Creutzfeldt-Jakob hastalığı, Rehabilitasyon

Introduction

Sporadic Creutzfeldt-Jakob disease (sCJD) is a rare transmissible neurodegenerative disorder with an invariably fatal outcome [1]. The disorder starts in the middle to late ages and results in rapidly progressive dementia, myoclonus and psychiatric disorders [2]. The clinical presentation of sCJD can be highly variable and overlap with other central nervous system disorders. In this article, we present an individual with difficulty in walking and balance and whose complaints were progressively advancing during rehabilitation.

Case presentation

A 78-year-old female presented to our outpatient department with a 5-months history of obliviousness, bilateral lower limb weakness, progressive difficulty in walking, balance disorder, urinary incontinence and weakness of the hands. There was no family history of neurological illnesses and her previous medical history included mild hypertension and gastritis. She had already been evaluated by neurology and neurosurgery departments. Biochemical and hematological investigations, cerebrospinal fluid examination, brain magnetic resonance imaging (MRI) and diffusion-weighted imaging (DWI) were unremarkable. Electroneuromyographic evaluation revealed mild peripheral neuropathy. MRI of the whole spine revealed narrowing of disc space at multiple levels with spinal cord compression at cervical level. The patient was transferred to our rehabilitation clinic, with a diagnosis of cervical spinal stenosis.

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At first presentation; the patient was conscious, slightly disoriented and had difficulty in cooperating. Her mimics were apathetic and she had titubation-like oscillations of the head, she also complained of obliviousness. In motor examination, upper and lower extremity strength was 4/5 except for bilateral hip flexion which was 3/5. There was diffuse numbness on the legs. Both upper and lower extremity deep tendon reflexes were increased, Hoffman's sign was bilateral positive and Babinski reflexes were bilaterally negligent. Her cognitive functions regressed rapidly including obliviousness. Her speech became dysarthric and she developed limb rigidity, dementia signs, spontaneous myoclonus and aggressive personality changes two weeks after initial presentation. She could not be mobilized without the support of a person. She was enrolled in the rehabilitation program. During her hospital stay, she showed progressive decline in gait and balance. At this stage, diagnostic laboratory investigation was unremarkable, including full blood count, liver and renal functional tests, glucose profiles, serum electrolytes, thyroid function tests, vitamin B12 and folate concentrations, creatine kinase, paraneoplastic antibodies, heavy metals, and tumor markers. The electroencephalography (EEG) and brain DWI were re-evaluated. DWI demonstrated symmetrical diffusion limitation seen in bilateral caudate nuclei and the putamens (Figure 1) and EEG showed widespread marked slowing of cerebral bioelectrical activity. Cerebrospinal fluid analysis was positive for 14.3.3 protein. The patient became completely dependent despite rehabilitation and was transferred to the neurology clinic with a diagnosis of probable sCJD.

We obtained written informed consent from the patient's legal representative for this report.

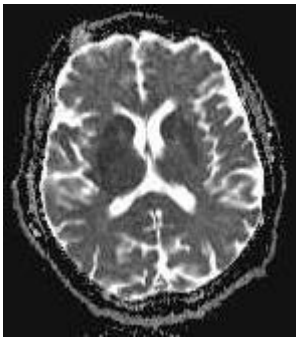

Figure 1: The brain diffusion-weighted images of the patient demonstrated symmetrical diffusion limitation in bilateral caudate nuclei and the putamens

Discussion

The most common human prion disease is sCJD, accounting for about 85% of all CJD, with an annual incidence of one person per million. The majority of the cases are sporadic while the remainder has a genetic component and early recognition is important in terms of genetic screening of the family. The median age of onset is 65 years, and the median duration of survival is four months [1]. It is due to the action of neurolytic pathogen proteins, called prions, which gradually damage central nervous system cells resulting in brain damage with distinct pathologic features [3]. The well-known clinical manifestations of sCJD are rapidly progressive dementia, myoclonus, and ataxia. At follow-up dementia with ataxia, personality changes and a variety of neurological and neuro-behavioral symptoms such as psychosis develop [4]. The diagnosis of sCJD is relatively straight forward when a patient with rapidly progressive dementia manifests myoclonus and periodic, synchronous, and generalized bi-/triphasic sharp-wave complexes on EEG [5]. Definitive diagnosis requires neuropathological examination; this is, however, no longer necessary because of the established WHO disease defining criteria [6] (Table 1) and a brain biopsy is discouraged unless required to exclude a treatable disease [1]. The most important reason for the initially misdiagnosis in our case was the lack of a clinically significant change in cognitive function at first presentation. In a case presented by Grant et al. [7], the 39-year-old patient was initially admitted with the diagnosis of spinal cord injury with weakness in the lower extremities and incontinence. Neuro-axis imaging studies failed to explain the symptoms and the patient's complaints were thought to have a large psychologic component. Cerebrospinal fluid analysis performed due to the development of neurological symptoms proximal to the presumed spinal cord injury level was suggestive of prion disease.

Table 1: World Health Organization clinical diagnostic criteria for sporadic Creutzfeldt-Jakob disease

Creutzfeldt-Jakob Disease
Progressive dementia
And at least two of the following four clinical features
Myoclonus
Visual or cerebellar disturbance
Pyramidal/extrapyramidal dysfunction
Akinetic mutism
And
A typical electroencephalogram during an illness of any duration and/or
A positive 14-3-3 CSF assay and a clinical duration to death <2 years
Routine investigations not suggestive of an alternative diagnosis

Brain DWI, assessment of 14-3-3 protein from cerebrospinal fluid and serial EEG recordings should be integrated into the diagnostic workup of sCJD [5]. In the literature, DWI is the most sensitive method in the diagnosis of sCJD. Its sensitivity is 96%, and the specificity is 93% [8]. The initially brain DWI was normal in our case which also played a role in the misdiagnosis. Shea et al. [9] described a 68-year-old male patient initially diagnosed with cervical myelopathy but who later developed rapidly progressive ataxia and was finally diagnosed with sCJD after extensive investigations. Similarly, initial DWI of the patient was unremarkable.

In our patient, the rapid and progressive course in walking difficulties, cognitive functions, progressive dementia, ataxia, myoclonus and personality changes suggested the possibility of the onset of sCJD. Laboratory findings for hypothyroidism, hypovitaminosis, tertiary syphilis and HIV infection were unremarkable. We did not consider dementia with Alzheimer's disease and Lewy bodies in differential diagnosis due to the rapid progression of dementia and cognitive functions. In addition, there was no remarkable history that suggested intoxication. Typical EEG, cerebrospinal fluid analysis and brain DWI findings in our patient suggested a probable sCJD. Biopsy/autopsy is required for a definite diagnosis.

In a sCJD case, reported by Gialanella et al. [3], the patient did not show any functional improvement despite rehabilitation. Similarly, functional improvement was not observed after rehabilitation in our case.

We conclude that in case of rapidly progressive dementia, ataxia and gait impairment, sCJD should always be considered even if unusual features are present and current diagnostic criteria are not completely fulfilled. sCJD is not a

treatable disease however diagnosis is important for palliative treatment of conditions such as pulses and myoclonus. Accurate diagnosis is also important to prevent the transmission of disease.

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