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Sporadic Creutzfeldt-Jakob Disease with Isolated Cerebellar Findings at Onset

İzole Serebellar Bulgular ile Başlangıç Gösteren Creutzfeldt-Jakob Hastalığı

Nurettin Yavuz, Yüksel Erdal, Ufuk Emre, Arife Çimen Atalar, Osman Özgür Yalın
Istanbul Training and Research Hospital, Clinic of Neurology, Istanbul, Turkey

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Anahtar Kelimeler: Creutzfeldt-Jacob hastalığı, serebellar bulgular, demans

Dear Editor,

A 53-year-old male patient was admitted to our hospital with symptoms of imbalance lasting 15 days. It was learned that his symptoms had increased within days and led to further deterioration in his walking. His medical history was normal except for hypertension. On neurologic examination, pursuit and saccadic eye movements were impaired, and cerebellar dysarthria was present. Muscle strength was normal. Cerebellar tests were bilaterally impaired, evident on the left. He had a wide-based gait and truncal ataxia. Tandem gait was impaired, and the Romberg test was positive. Cranial magnetic resonance imaging (MRI) revealed gyral signal increase at the bilateral head of the putamen and caudate nucleus, and at bilateral cerebral cortical areas and bilateral cerebellum, more evident at the right parieto-occipital region in diffusion-weighted and fluid-attenuated inversion recovery sequences (Figures 1, 2). Cerebrospinal fluid (CSF) biochemistry and cell count were within normal limits. During follow-up, a significant worsening of the cerebellar findings was observed within two weeks. He had severe dysarthria, and his truncal ataxia became prominent. There was no myoclonus. Akinetic mutism developed three weeks after the onset of the symptoms. The patient, fed with a nasogastric tube, became bedridden. Diffuse slow background activity was observed in the electroencephalography (EEG). Periodic sharp wave complexes

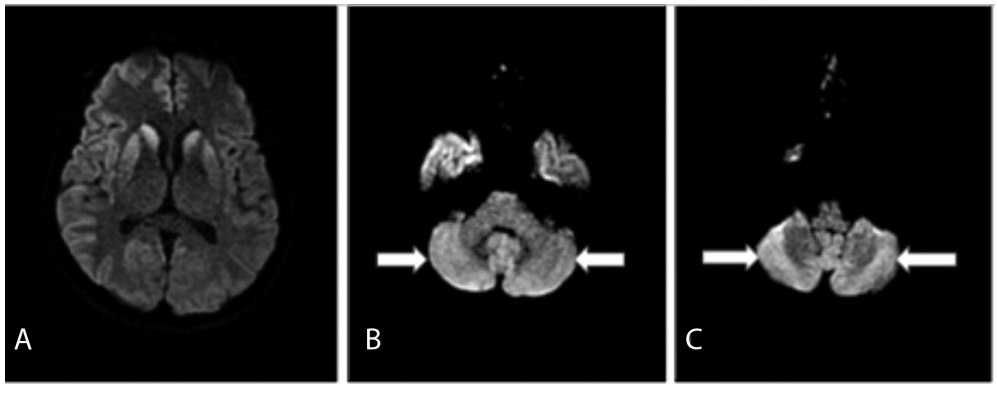

Figure 1. Signal increase in the bilateral cerebral cortical areas prominent in the bilateral basal ganglia and right hemisphere (A) and cerebellar cortical (B and C) areas on diffusion-weighted imaging.

Address for Correspondence/Yazışma Adresi: Nurettin Yavuz MD, Istanbul Training and Research Hospital, Clinic of Neurology, Istanbul, Turkey
Phone: +90 542 279 56 03 E-mail: nurettinyavuz85@hotmail.com ORCID ID: orcid.org/0000-0002-8670-2438
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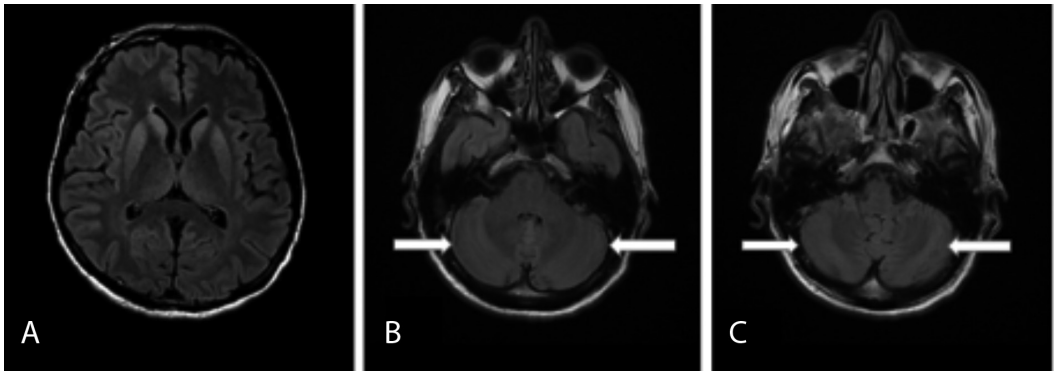

Figure 2. Signal increase in the bilateral cerebral cortical areas prominent in the bilateral basal ganglia and right hemisphere (A) and cerebellar cortical (B and C) areas on FLAIR sequence.

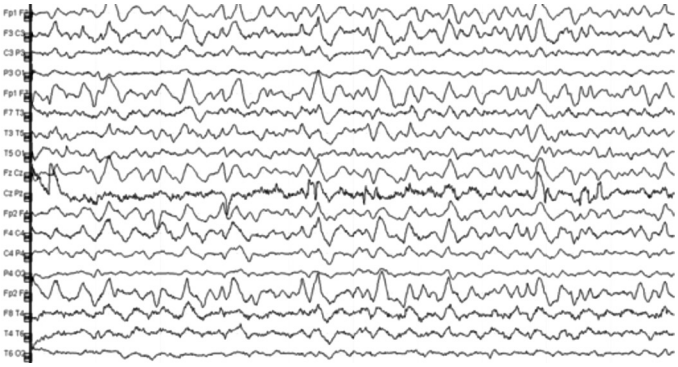

Figure 3. Sharp slow wave paroxysms in the electroencephalography

appeared in follow-up EEGs three weeks later (Figure 3). Sporadic Creutzfeldt-Jakob disease (CJD) was diagnosed considering the clinical, imaging, EEG, and CSF findings of the patient, along with CSF 14-3-3 positivity. During the first month of hospitalization, the patient developed aspiration pneumonia. He died on the 15th day of intensive care follow-up.

CJD is a neurodegenerative disease caused by prions and has a rapidly progressive course. Sporadic CJD (sCJD) is the most common subtype and is mortal (1). The classic clinical findings of sCJD are typical rapid progressive dementia, cerebellar findings, myoclonus, and akinetic mutism in the later period; however, a wide variety of clinical features can be observed at the onset of the disease (2). Cerebellar findings are common in sCJD and occur in the early period. In some cases, sCJD may begin with isolated cerebellar clinical findings, also called the Brownell-Oppenheimer variant (3). In our case, there were only cerebellar findings at the beginning, but dementia and akinetic mutism were added to the clinical picture later, but myoclonus was not observed during the follow-up.

Several cases have been reported in the literature, beginning with isolated clinical findings such as visual findings, speech disorders, cerebellar findings and monoparesis. In a study, focal findings were initially detected in 80% of cases (2). In a study conducted by Appleby et al. (4), cerebellar findings were reported as 53% and were reported as the most common initial finding in 22%. In an autopsy series performed by Cooper et al. (2), clinical onset with isolated cerebellar findings were observed in 5%.

Diagnosis may be delayed or overlooked if there are no EEG and MRI changes with diagnostic features in patients with isolated focal and atypical onset (2). MRI findings are particularly important for both excluding other diseases and assessing patients who are suspected of having focal or atypical onset by showing features that are typical of sCJD from the early stage of the disease (5). In our patient, who presented with isolated clinical findings, the clinical findings at onset did not suggest CJD, but the patient was assessed in this respect due to diffusion MRI findings suggesting sCJD.

CJD can be considered in the differential diagnosis of patients presenting with acute-subacute cerebellar findings. In particular, imaging modalities have great prospects for early diagnosis in suspect cases that may lead to diagnostic confusion.

Ethics

Informed Consent: Consent form was filled out by all participants.

Peer-review: Internally peer-reviewed.

Authorship Contributions

Medical Practices: N.Y., Concept: N.Y., Y.E., Design: N.Y., Y.E., Data Collection or Processing: N.Y., Analysis or Interpretation: U.E., A.Ç.A., O.Ö.Y., Literature Search: N.Y., Y.E., Writing: N.Y.

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