OLGU SUNUMU/CASE REPORT

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Sporadic Creutzfeldt-Jakobs Disease with Hypothermic Progress: A Rare Case Report

Hipotermik Seyreden Sporadik Creutzfeldt-Jakobs Hastalığı: Nadir Bir Vaka

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ABSTRACT

Sporadic Creutzfeldt-Jacobs Disease (CJH) is spongioform encephalopathy caused by prion that should be considered in cases of dementia. It starts with dementia and neuropsychiatric findings in the middle age. Disease symptoms are not characteristic and can be confused with neurological and psychiatric diseases. A 49-year-old male patient was brought to the emergency room with complaints of not speaking, inability to move, memory loss and hearing loss, gait difficulty, intermittent agitated behavior, cold sweating, and vomiting. Her skin looked pale and cold. His temperature was 35 ° C. He had anxiety. The disease is diagnosed by clinical, laboratory, radiological examination, and EEG. Protein 14.3.3, a protease inhibitor protein released from neurons into the cerebrospinal fluid, is a biochemical marker with high sensitivity and specificity used in diagnosis. In radiological imaging, enlargement of the sulci, cerebral and cerebellar atrophy are helpful in the diagnosis of magnetic resonance imaging (MRI). In this study, we examined a rare, rapid and progressive, mortal, neurodegenerative prion disease, a sporadic case of Creutzfeldt-Jakob disease (CJD).

Keywords: Creutzfeldt-Jakobs Disease, dementia, hypothermi

ÖZ

Sporadik Creutzfeldt-Jacobs Hastalığı (CJH), demans vakalarında düşünülmesi gereken, prionun neden olduğu spongioform ensefalopatidir. Orta yaşlarda demans ve nöropsikiyatrik bulgularla başlar. Hastalık belirtileri karakteristik değildir ve nörolojik ve psikiyatrik hastalıklarla karıştırılabilir. 49 yaşında erkek hasta konuşamama, hareket edememe, hafıza kaybı ve işitme kaybı, yürüme güçlüğü, aralıklı ajite davranış, soğuk terleme ve kusma şikayetleri ile acil servise getirildi. Teni solgun ve soğuk görünüyordu. Ateşi 35 °C idi. Kaygısı vardı. Hastalık klinik, laboratuvar, radyolojik inceleme ve EEG ile teşhis edilir. Nöronlardan beyin omurilik sıvısına salınan bir proteaz inhibitörü protein olan Protein 14.3.3, tanıda kullanılan yüksek duyarlılık ve özgüllüğe sahip biyokimyasal bir belirteçtir. Radyolojik görüntülemede sulkus, serebral ve serebellar atrofinin genişlemesi manyetik rezonans görüntüleme (MRI) tanısında yardımcıdır. Bu çalışmada, nadir, hızlı ve ilerleyici, ölümcül, nörodejeneratif bir prion hastalığı, sporadik bir Creutzfeldt-Jakob hastalığı (CJD) vakasını inceledik.

Anahtar Kelimeler: Creutzfeldt-Jakob hastalığı, demans, hipotermi

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INTRODUCTION

Creutzfeldt Jacob's Disease (CJD) was first described by Creutzfeldt and Jacobs in the 1920s, and it has a clinical manifestation of rapidly progressive dementia, myoclonus, ataxia, visual disturbances, extrapyramidal and pyramidal involvement, and akinetic mutism (1). Psychiatric symptoms such as depression, psychosis, anxiety, cognitive impairment, and irritability may occur in 80-90% of the patients. It has four forms: sporadic, variant, familial and iatrogenic. Sporadic form occurs with somatic gene mutation or spontaneous prion protein transformation. This form constitutes 85% of the cases. It is seen equally in men and women. Its incidence is approximately one in a million per year (2). In this study we would like to emphasize a rare, rapid and progressive, mortal, neurodegenerative prion disease, a sporadic case of Creutzfeldt-Jakob disease (CJD).

CASE REPORT

49-year-old male patient. Lack of appetite, cold sweating, vomiting, not speaking for the last two days, inability to move, memory loss and hearing loss for the last 20 days, walking difficulties, intermittent agitated behavior, rapid growth in the last three months. The patient, who was diagnosed with myocardial infarction five months ago was stented for coronary arteria in the coronary intensive care unit of our hospital. The patient who was evaluated in the emergency room was conscious, non-oriented, non-cooperative, and there was no PR and MIB. Her temperature was 36 ° C ↓ and he was hypothermic. He did not follow orders except simple commands. Pupillary isochoric, IR (+ / +). Blood pressure: 90/50 mm-hg, Pulse: 92 / min, S1, S2 were arrhythmic. His skin was pale and cold, marble-like, and his gaze looked dull. Lung sounds decreased in basal. Hepatomegaly and splenomegaly were present in the abdomen. In laboratory tests, Hbs Ag: negative, Anti Hbs: negative, Anti HCV: negative, Anti HIV: negative, syphilis ELISA: negative, Ebstein Barr Virus (EBV) VCA IgM, IgG negative. White blood cell (WBC): 3800, neutrophile (NE) 89 %, HGB: 7.4, Plathelet: 60000, C-Reactive protein (CRP): 246, urine test: erythrocyte: 8, leukocyte: 3, UREA: 64, creatinine: 1.38, aspartat aminotransferase (AST): 89, alanine aminotransferase (ALT): 14, Gamma glutamyl transferase (GGT): 92, Alkaline phosphatase (ALP) : 165, B.total: 1.58, PT: 19.5, INR: 1.65, APTT: 45. Abdominal tomography: hepatomegaly 192 mm, splenomegaly 162 mm with free fluid between the bowel loops, thorax CT: marked 34 mm pleural effusion on the right and right lung There was pneumonic consolidation. Cerebral and cerebellar cortical-subcortical atrophy, enlarged cerebral sulci, cerebellar folia and ventricular system secondary to atrophy, mucosal thickening in the right maxillary sinus on brain MRI. MR venography: Right transfers, sigmoid sinus, internal jugular vein were hypoplastic, and compensatory on the left. Meropenem 3x1 IV, vancomycin 2x1 IV, acyclovir 3x750 mg IV treatments were started. The patient, who was arrested in the ward for a few days, was followed up in the post-CPR intensive care unit. There was no reproduction in blood, urine and TAK cultures taken during follow-up. Lumbar puncture (LP) was performed on the 14th day of his hospitalization. Cerebrospinal fluid (CSF) was transparent, pressure increased. No leukocyte erythrocyte was seen in the cell count. Nonspecific CSF culture was not grown. CSF, Erlich Ziehl Nilsen (EZN) was negative, CSF mycobacterium Polimerase Change Reaction (PCR) and HSV DNA PCR were negative. Glucose in CSF biochemistry: 81 mg / dl (40-70 mg / dl), sour: 198 mg / dl protein: 101 (15-45 mg / dl), LDH: 672 (0-20 U / L), Cl: 125 (98-107 mEq / L). A diagnosis of CJH was made with a positive result of the protein 14.3.3 test from the CSF. WBC: 3500 NE: 32%, HGB: 10.6, PLT: 59000, Urea: 139, creatinine: 1.35, Aspartate aminotransferase (AST): 377, Alanine aminotransferase (ALT): 160, Lactat dehidrogenase (LDH): 667, Gamma glutamyl transferase (GGT): 285, Alkaline phosphatase (ALP): 243, T .bil: 5.25, D.bil: 3.46, C-Reactive protein (CRP): 325. 21st days of hospitalization he was dead.

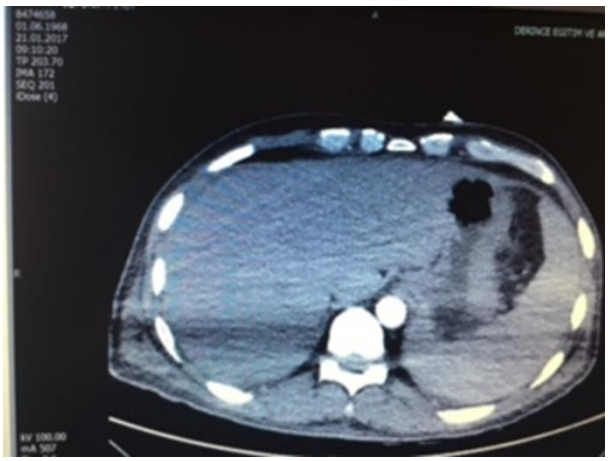

Figure 1. Abdominal CT: Contrast-enhanced computed tomography, hepatomegaly, splenomegaly on abdominal examination

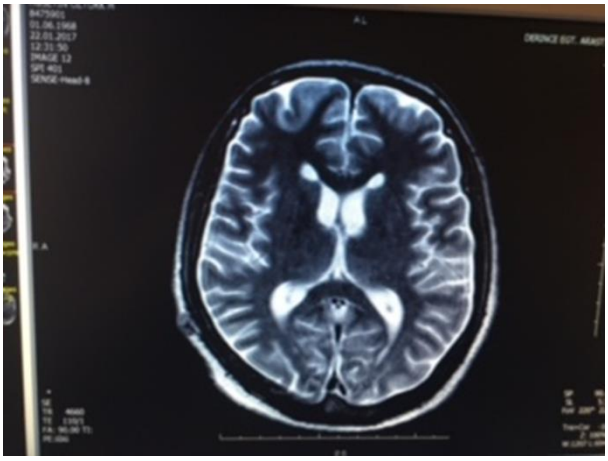

Figure 2. Brain Contrast-enhanced MR: T2 hyperintense, cortical subcortical atrophy, ventricular enlargement

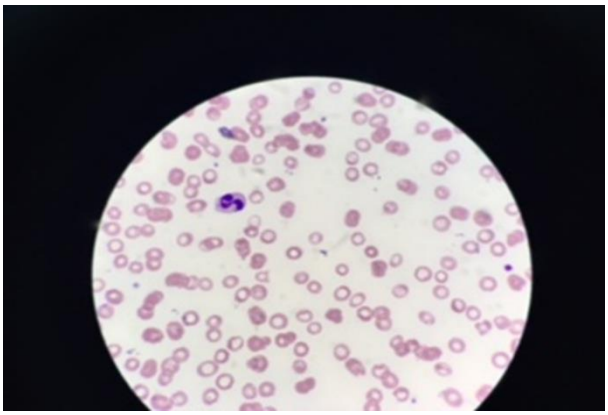

Figure 3. Peripheral smear examination: Hypochromic stained, anisocytosis erythrocytes

CONCLUSION

Creutzfeldt Jacob's Disease is the most common neuroegenerative disease among spongioform encephalopathies, with variant, familial, iatrogenic and sporadic varieties. Although the sporadic form is most common, it occurs with spontaneous prion protein transformation or somatic gene mutation. It is usually seen in the sixth decade and women and men are affected equally. Myoclonus, ataxia and rapidly progressive dementia, rigidity, myoclonus, and akinetic mutism are the main findings. Our patient also had these findings (3,4). Among the laboratory tests, the increase in protein 14.3.3 in the CSF is 96%, its sensitivity is 99% in the diagnosis of sporadic CJH. This test was also positive in our patient. Typical periodic spike wave appearance in EEG (electroencephalogram) is significant in the presence of clinical findings. In MR (Magnetic Resonance) examination, prion protein accumulation in the brain is associated with tissue damage, brain atrophy, cortical signal increase and thalamus and basal ganglion involvement sensitivity is 71%, specificity is 91%. With the PRNP (prion protein gene) test, methionine, valine polymorphism and gene mutation can be determined in 129 codons of the prion protein(5,6). In a study conducted with 248 patients, 64% of sporadic CJH patients had agitation, 45% hallucinations, 50% anxiety and 37% depression. Findings in our case were also consistent with this study. Histopathological examination of postmortem brain biopsy and demonstration of prion protein by immunohistochemical staining are important in definitive diagnosis (7). Recently 'the real-time quaking-induced conversion' test (RT-QuIC) diagnosed with JCD, the sensitivity and specificity of PrPSc detection in CSF or mucosal brushing sample is high (10). Although the average life span of the disease is four months, 85% of the patients die within a year.

We emphasized a rare, rapid and progressive, mortal, neurodegenerative prion disease, a sporadic case of Creutzfeldt-Jakob disease (CJD). Alzheimers disease, central nervous system infections, autoimmune encephalitis, frontotemporal dementia, toxic

and metabolic diseases should be kept in mind in the differential diagnosis. CJD should be considered in the differential diagnosis of cases with neurological findings (8,9,11). Sporadic JCD disease, which is rarely seen in rapidly progressive dementia cases, should be kept in mind in the differential diagnosis.

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