Diagnostic pitfalls in Creutzfeldt-Jakob disease

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Background and aims: Creutzfeldt-Jakob disease (CJD) is a very rare neurodegenerative disease of the brain. One of the main characteristics of CJD is the rapidly progressive dementia. The clinical tableau consists in: rapidly progressive dementia, myoclonic jerks and a variety of neurological abnormalities. The particularity of our case is the lack of dementia which raised the difficulty level in establishing a diagnosis. Case Description: Our patient is a 55 year old female who suddenly presented gait disturbance and left limb ataxia. She was hospitalized for what was supposed to be a stroke, performed a CT without any abnormalities. Almost immediately she developed myoclonic contractions on her left limb. We found: paraparesis, myoclonia on her left limbs, bilateral horizontal nystagmus, dysmetria, dysarthria and bilateral Noica sign and all her intellectual functions were preserved. The laboratory tests were normal, the cerebral MRI showed a hyperintensity in T2 and FLAIR -weighted images of the basal ganglia and right occipital cortex. The EEG was uggestive for CJD. The spinal fluid was positive for the “14-3-3” protein. At this point the diagnosis of CJD was established. The seizures gradually became generalized in spite of the maximal antiepileptic treatment. She became comatose due to this state, was intubated and died within 2 months since onset. Conclusion: Till the moment on which our patient became comatose she didn’t present any signs of dementia. We shouldn’t disregard the CJD diagnosis in the absence of cognitive impairment.