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The role of brain FDG-PET in the diagnosis of Creutzfeldt-Jakob disease (CJD): A case report

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Objective: Creutzfeldt-Jakob disease (CJD) is a rare and fatal neurodegenerative disease, whose main clinical features are rapidly progressive mental deterioration, myoclonus, ataxia, and visual disturbances. Its clinical diagnosis can be challenging, and definitive confirmation is obtained by brain biopsy. However, its role has been established by positive 14-3-3 (CSF) cerebrospinal fluid essay, supportive electroencephalogram (EEG) findings and magnetic resonance imaging (MRI). Besides, brain FDG-PET shows high specificity and even higher sensitivity than MRI in improving diagnostic accuracy, especially in the early stages of the disease.

Materials & Methods: JM, male patient, 79 years old, was first evaluated in October 2017 because of subjective gait impairment, initial word finding difficulties, sleep disturbances, emotional liability and anxiety. He had no previous pathological history and was not on medication. The neurological examination was normal. At that time we began treatment with benzodiazepines (BDZ) and selective serotonin reuptake inhibitor (SSRI). After a month he presented again with progressive aphasia, ataxia and myoclonus in the left hemisoma. He was then admitted in our department to undergo the investigations for prion disease.

Results: MRI in our case was not conclusive, failing to show the typical signal abnormalities in caudate nucleus and/or putamen on diffusion-weighted imaging (DWI). The electroencephalogram (EEG) showed only diffuse electrical brain abnormalities. Therefore, we performed a brain FDG-PET scan, which showed a clear hypo-metabolic pattern in the left caudate and in the left parietal-temporal-occipital cortex; a mild hypo-metabolism in the left frontal and right occipital regions. These findings, when present, are suggestive of CJD. In the next two months, the patient progressively developed an akinetic mutism which inexorably worsened until he expired four months after the initial onset of the symptoms. The CSF results came after his death showing positive 14-3-3 results, high total tau levels (2131 pg/ml) and positive RT-QuIC which confirmed the diagnosis of CDJ.

Conclusions: While brain biopsy remains the only confirmatory diagnosis of CJD, a role has been established for CSF examination, and MRI findings. Brain FDG-PET is another non-invasive, easy performing and rapid investigation which has proven to be of great diagnostic support when other investigations are not available or inconclusive.

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