CAS CLINIQUE

MRI and FDG PET/CT findings in a case of probable Heidenhain variant Creutzfeldt-Jakob disease

Aspects en IRM et TEP/TDM au FDG d’un cas de variant Heidenhain de maladie de Creutzfeldt-Jakob probable


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Résumé La maladie de Creutzfeldt-Jakob (MCJ) est une maladie neurodégénérative causée par l’accumulation d’un isoforme pathogène de la protéine du prion dans les neurones, responsable d’un tableau de démence subaigu. Le variant Heidenhain est une forme atypique de la MCJ pour laquelle les signes visuels sont au premier plan. Les auteurs rapportent le cas d’un variant Heidenhain de MCJ probable chez un patient de 65 ans, avec présentation des...
Introduction

Sporadic Creutzfeldt-Jakob disease (sCJD) is an infrequent cause of rapidly progressive dementia. Its major symptoms are cognitive dysfunction, myoclonus and pyramidal syndrome. The Heidenhain variant is a rare presentation of sCJD in which visual disturbances such as blurred vision, hallucinations or cortical blindness are at the forefront [1,2].

We report on the case of a probable Heidenhain variant sCJD on the basis of findings from electroencephalography (EEG), fluid-attenuated inversion recovery (FLAIR) imaging and diffusion-weighted images (DWI) on MRI, as well as, on 18F-fluorodeoxyglucose positron emission tomography/computed tomography (18F-FDG PET/CT) of the cortical parieto-occipital lesions.

Case report

A 65-year-old male patient was referred to the neurology department because of rapidly worsening dementia. He had no previous history of neurological disorder. The patient presented with recent subacute dementia with anterograde amnesic disturbances and hallucinations, but without fever. Neuropsychological examination revealed disorientation, psychomotor slowdown and anosognosia.

The physical examination showed no somatic abnormality and the initial EEG, performed at the beginning of the symptoms, was normal. One week later, a second EEG (not shown) demonstrated marked slowing of basic activity and a generalized synchronous slow discharge of periodic sharp wave complexes (PSWC). A state of hypovoltage was also observed in the right occipital area.

The patient underwent the following work-up: laboratory assays; contrast-enhanced CT; MRI (1.5-T) and FDG PET/CT. Also, serum tests for Mycobacterium tuberculosis, Borrelia burgdorferi, herpesvirus, HIV and syphilis (Treponema pallidum hemagglutination [TPHA] and VDRL) were all negative.

Analysis of cerebrospinal fluid (CSF) showed one per millimeter cube white cell, normal glucose and protein levels but no 14-3-3 protein.

The contrast-enhanced CT scan (not shown) was normal. MRI axial FLAIR-weighted images showed bilateral hyperintensities in the cortical ribbon of the parieto-occipital area, with right dominance. No abnormality was seen in the basal ganglia (Fig. 1A). DWI showed hyperintensities in the parieto-occipital cortices (Fig. 1B). The apparent diffusion coefficient (ADC) was decreased in these cortical lesions. Moreover, areas with DWI hyperintensities were in accord with those seen on FLAIR-weighted images (Fig. 1A and B). FDG PET/CT was performed because, as paraneoplastic syndrome was initially evoked, a primitive tumor was
reveals lesions involving the cortices and/or basal ganglia [3]. MRI is the imaging method of choice in sCJD and often and EEG findings were consistent with the published criteria this patient most probably had sCJD as his clinical symptoms According to the French and European diagnostic criteria, Discussion 18F-FDG (Fig. 2) and showed decreased uptake in the pari- etooccipital areas — the same areas that appeared abnormal on MRI — and in the right frontal-lobe cortices. However, as no focus of high FDG uptake was seen in the rest of the body, paraneoplastic syndrome was ruled out. In fact, the clinical presentation of rapidly worsening dementia associated with visual disturbances and certain EEG findings were strongly evocative of Heidenhain variant sCJD, according to the criteria for CJD diagnosis [3,4]. Moreover, the cortical ribbon involvement seen on the brain MRI scans also supported this diagnosis. In the months following hospitalization, the patient’s condition worsened rapidly and he eventually died three months later. No autopsy was performed as the clinical symptoms and EEG findings were sufficient for a diagnosis of probable sCJD and also because of the difficulty of such a procedure, which is now reserved only for suspected familial cases.

Discussion

According to the French and European diagnostic criteria, this patient most probably had sCJD as his clinical symptoms and EEG findings were consistent with the published criteria [3]. MRI is the imaging method of choice in sCJD and often reveals lesions involving the cortices and/or basal ganglia [5,6,7,8]. These lesions are characterized by increased signals on both FLAIR and DWI sequences. Hyperintensities on DWI are associated with a decreased ADC and are another strong argument in favor of such a diagnosis, as the diagnostic gamut of these changes is limited. As found in several studies, MRI changes are correlated with pathological findings that involve spongiform change, astrocytic gliosis and neuronal loss [9,10]. Indeed, the association of FLAIR-weighted images and DWI lesions is 91% sensitive, 95% specific and 94% accurate for a diagnosis of CJD [11]. In addition, although positive findings on MRI are not a criterion for a diagnosis of sCJD, it can help to establish the diagnosis. Differential diagnoses for cortical hyperintensities on FLAIR and DWI are mainly cerebral hypoxia, cortical hypoxia and postictal changes. FLAIR and DWI hypersignals in the basal ganglia associated with subacute dementia may also be seen in carbon monoxide (CO) intoxication, Leigh’s disease and Wilson’s disease [12].

FDG PET/CT may be a useful tool for early diagnosis of CJD as it may reveal lowered cellular glucose transport and metabolism in the cortices (as in the present case), cerebellum and/or basal ganglia [13]. Decreased FDG uptake may well be related to neuronal damage, leading to glucose hypometabolism [14,9]. FDG PET/CT may also help to eliminate differential diagnoses of CJD such as paraneoplastic encephalopathy, in which hypometabolism is predominantly seen in the temporal lobes and primitive neoplasm, which may be uncovered in whole-body images. Low uptake in the frontoparietal areas of the brain, as found in sCJD, may also be seen in other dementias such as Alzheimer’s disease [15]. To our knowledge, only two cases of FDG PET/CT findings have been previously reported for Heidenhain variant sCJD [13,16]. In those two cases, as in the present one, hypometabolism was seen in the parietooccipital areas at the same locations as the FLAIR and DWI hyperintense signals on MRI. Moreover, it is worthwhile noting that, on EEG findings, a state of hypovoltage was observed in those same parietooccipital areas, especially on the right. This state could be related to severe neuronal loss (as suggested by the strong FLAIR and DWI hyperintensities seen on MRI and the low FDG uptake on PET/CT), while synchronous slow discharge in the right frontal lobe on EEG could be associated with less severe neuronal damage or loss. Moreover, as PET/CT hypometabolism and DWI/FLAIR signal abnormalities did not match for frontal cortical lesions, this may be an indication of a greater sensitivity of PET/CT to identify early lesions. In contrast to previously reported cases, the 14-3-3 protein was, in the present case, negative in the CSF. This serves to emphasize that imaging findings (MRI and FDG PET/CT) may be of great value in the diagnosis of sCJD, especially in cases where a major criterion for a positive diagnosis of sCJD, such as the presence of 14-3-3 protein in CSF, is not found.

Conclusion

Our case report presents EEG, MRI and 18F-FDG PET/CT findings of probable Heidenhain variant sCJD. Most of the abnormalities on 18F-FDG PET/CT scans are consistent with...
those found on DWI and FLAIR images and are believed to correspond with areas of neuronal loss. Other foci of low uptake on PET/CT images that do not match MRI abnormal findings might be indicative of early cortical lesions. The present case suggests that 18F-FDG PET/CT may be useful for early sCJD diagnosis and help to eliminate other differential diagnoses of subacute dementia.

References