Print ISSN 1738-1495 / On-line ISSN 2384-0757
Dement Neurocogn Disord 2017;16(3):91-93 / https://doi.org/10.12779/dnd.2017.16.3.91

LETTER TO THE EDITOR

Familial Creutzfeldt-Jakob Disease with M232R Mutation Progressed Slowly like Alzheimer's Disease

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Dear Editor,

Creutzfeldt-Jakob disease (CJD) is a typically fatal neurodegenerative disease, and 90% of CJD patients die within 1 year of diagnosis. Of all CJD cases, 10−15% are autosomal dominant disorders of the prion protein gene (*PRNP*) in chromosome 20, classified as genetic or familial CJD.

Familial CJD has an younger onset and has a lengthier disease progression than sporadic CJD.[1] In the past, clinical patterns of CJD with M232R mutation (CJD232) were like sporadic CJD. However, CJD232 cases that reveal longer clinical courses and periodic sharp and wave complexes distinctively found in an eletroencephalography (EEG) are not observed have been recently reported.[2] The authors report cases of patients diagnosed with familial CJD revealed slow progress like Alzheimer's disease.

A 62-year-old male patient visited our hospital because he had gait disturbance 2 years ago and symptoms of memory deterioration, etc. occurred 1 year ago. In his medical history,

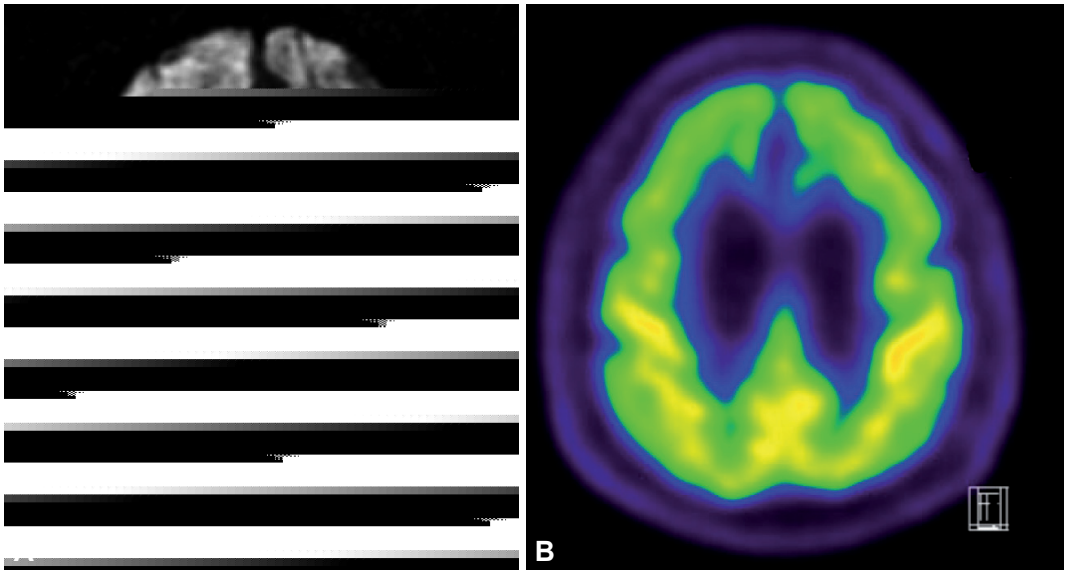

Fig. 1. Initial DWI and 18F-fluorodeoxyglucose PET CT. A: DWI demonstrates cortical high-signals. B: PET study shows diffuse hypometabolism of bilateral frontoparietal lobes. DWI: diffusion weighted image, PET: positron emission tomography.

Received: August 21, 2017 **Revised:** September 15, 2017 **Accepted:** September 15, 2017

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tial symptom to akinetic mutism was 3.1±1.5 months in the former type and 20.6±4.4 months in the latter type. Regarding mean time to reveal myoclonus, it took 2.4±1.8 months in the rapid type and 15.3±12.3 months in the slow type. Two different phenotypes were revealed in patients with accurately identical genotype. There were some cases that dementia with Lewy bodies was identified in the result of the final pathological autopsy of patients suspected of CJD with M232R mutation.[2]

The patient in the current case was assumed as Alzheimer's disease because he was 65 years old with cognitive deterioration that progressed 1 year ago at the first hospitalization and the patient did not have abnormal findings in psychataxia, myoclonus, and EEG. As CJD was suspected due to the lesion with high-signal intensity in the diffusion-weighted MRI, identified accidentally, he received cerebrospinal fluid examination and was finally diagnosed with familial CJD. However, he is now revealing slower clinical progress than the slow type of patients with CJD232 reported so far, and his image findings revealed no changes for 26 months, that is exceptional compared to the progress period published in other literature. In the future, it will be necessary to conduct more studies to identify if M232R mutation contributes to familial CJD, or studies on variation factors of Alzheimer's disease.

Conflicts of Interest

The authors have no financial conflicts of interest.

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