



Imaging Findings of Familial Dementia with a Tau R406W Mutation

Junji KOMATSU¹, Kenjiro ONO¹, Daisuke YANASE¹, Miharu SAMURAKI¹, Keisuke SHIMA¹, RYOZO KUWANO²,
Ichiro MATSUNARI³ and Masahito YAMADA¹

¹Department of Neurology and Neurobiology of Aging, Kanazawa University Graduate School of Medical Science;

²Department of Molecular Genetics, Center for Bioresources, Brain Research Institute, Niigata University;

³The Medical and Pharmacological Research Center Foundation, Hakui, Japan

A 67-year-old man presented with memory impairment progressing during 1 year. His two brothers and one sister had dementia. Except for memory impairment, he had no abnormal neurological findings. Brain MRI showed mild frontotemporal atrophy involving hippocampus (Fig. 1). Fluorine-18 fluorodeoxyglucose positron emission tomography (FDG PET) revealed marked hypometabolism in the temporal poles as well as mild hypometabolism in the right striatum and posterior cingulate gyri even after correction for atrophy (1) (Fig. 2). Carbon-11 Pittsburgh Compound B PET showed no retention in the cortex. A R406W mutation was identified in exon 13 of the tau gene. Hypometabolism in the posterior cingulate gyri is frequently observed in AD, but it was also recently described in frontotemporal dementia (FTD) patients (3). The clinical phenotype of FTD with this mutation is similar to that of Alzheimer's disease (AD). FDG PET could help in the differential diagnosis.

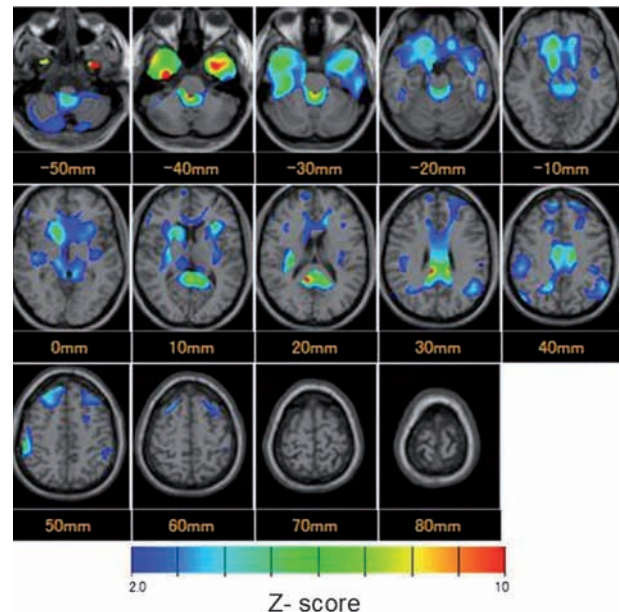


FIG. 2. — FDG PET z-score maps, displayed by overlaying on the anatomically standardized MRI image of healthy volunteers, showed remarkable reduction of the cerebral glucose metabolism in the bilateral temporal poles and mild reduction in the right striatum and posterior cingulate gyri.

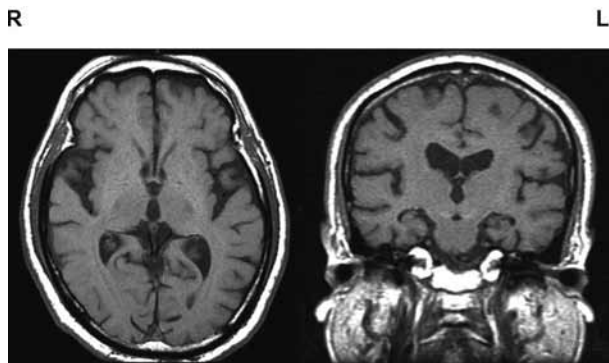


FIG. 1. — Brain MRI showed mild atrophy with mild frontotemporal atrophy. The MRI findings were not inconsistent with Alzheimer's disease.

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Pr. Masahito Yamada, M.D., Ph.D.,
Department of Neurology and
Neurobiology of Aging,
Kanazawa University Graduate School of
Medical Science,
13-1, Takara-machi,
Kanazawa 920-8640 (Japan).
E-mail: m-yamada@med.kanazawa-u.ac.jp