

Teaching NeuroImages: Cerebral cortex swelling in Creutzfeldt-Jakob disease with V180I mutation

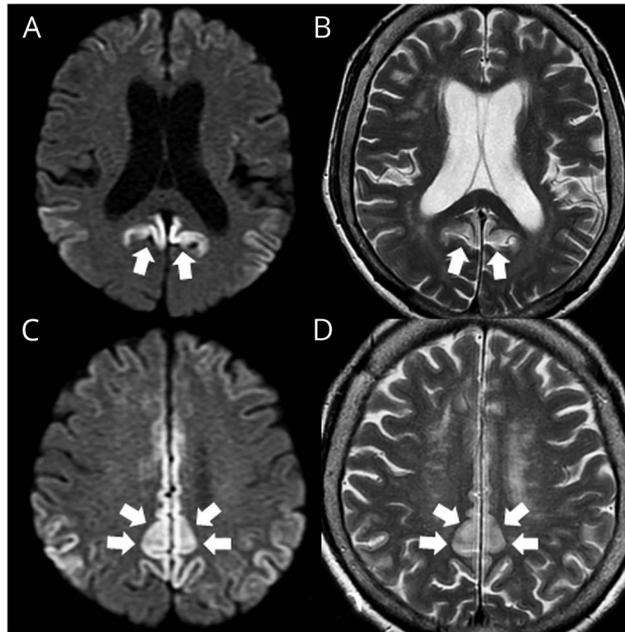
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Figure Brain MRI without contrast



Brain MRI at 6 months after onset reveals increased signal intensity in the cerebral cortices with swelling on diffusion-weighted images (A, C) and T2-weighted images (B, D) (arrows).

A 74-year-old woman developed amnesia that rapidly progressed over 6 months. She exhibited no apparent neurologic abnormalities, except for cognitive decline. Mini-Mental State Examination score was 20/30, mainly involving orientation and recent memory. EEG revealed no periodic synchronous discharge. CSF analysis was negative for 14-3-3 protein. Brain MRI revealed abnormal signals in the cerebral cortices (cortical ribboning) with swelling (figure). Analysis of the prion protein gene (*PRNP*) revealed V180I mutation. Premortem diagnosis of Creutzfeldt-Jakob disease with V180I mutation, especially in early stage, merely based on clinical features is difficult.^{1,2} MRI revealed cortical swelling that is associated with V180I mutation and prompts genetic testing.^{1,2}

Author contributions

Atsuhiko Sugiyama: performing patient clinical assessment, drafting the manuscript, and creating the figure. Minako Beppu: performing genetic counseling and providing assistance in patient clinical assessment. Satoshi Kuwabara: editing the manuscript.

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Disclosure

The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

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