Reviewer's report

Title: Familial Frontotemporal Dementia with Neuronal Intranuclear Inclusions is not a Polyglutamine Expansion Disease

Version: 1 Date: 10 August 2006
Reviewer: Mitsunori Yamada

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General
Mackenzie et al. analyze DNA samples, which were obtained from patients with familial frontotemporal dementia (FTD) with ubiquitinated neuronal intranuclear inclusions (NIIs), for CAG/CAA repeat expansions in polyglutamine-encoding genes, and find no significant repeat expansions in the genes examined. The authors also report negative immunohistochemical results of the intranuclear inclusions for expanded polyglutamine stretches. They conclude that familial FTD with ubiquitinated NIIs is not a polyglutamine expansion disease.
This is the third report by the same authors concerning FTD with NII, and is a carefully constructed genetic analysis. From a morphological view point, NIIs found in familial FTD patients are different from those seen in patients with polyglutamine diseases, and the results are somewhat predictable. The negative immunohistochemical result of NIIs for expanded polyglutamine stretches has been reported in a family linked to chromosome 17 by the same authors (#11 in Reference).

Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)
1. Page 5: It is not clear whether patients examined in the present study have been included in the authors' previous publications (#11 and 17 in Reference).

Discretionary Revisions (which the author can choose to ignore)

What next?: Accept after minor essential revisions
Level of interest: An article whose findings are important to those with closely related research interests
Quality of written English: Acceptable
Statistical review: No
Declaration of competing interests:
I declare that I have no competing interest.