Phenotype associated with APP duplication in five families

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Résumé en anglais	Different duplications of the APP locus have been identified in five families with autosomal dominant early onset Alzheimer's disease (ADEOAD) and Abeta-related cerebral amyloid angiopathy (CAA). This study describes the phenotype of this new entity. Clinical, neuropsychological, imagery and neuropathological data were reviewed. The phenotype was not dependent on the size of the duplication and there was no clinical feature of Down's syndrome. Dementia was observed in all cases; intracerebral haemorrhage (ICH) was reported in 6 (26%) and seizures occurred in 12 (57%) of 21 patients. Age of onset of dementia ranged from 42 to 59 years, ICH from 53 to 64 years and age at death from 46 to 75 years. The neuropathological findings in five cases demonstrated Alzheimer's disease and severe CAA lesions that were reminiscent from those reported in brains of Down's syndrome patients. A striking feature consisted in intraneuronal Abetax-40 accumulation located in the granular cell layer of the dentate gyrus and in the pyramidal cell layer of the Ammon's horn.
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