

Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy

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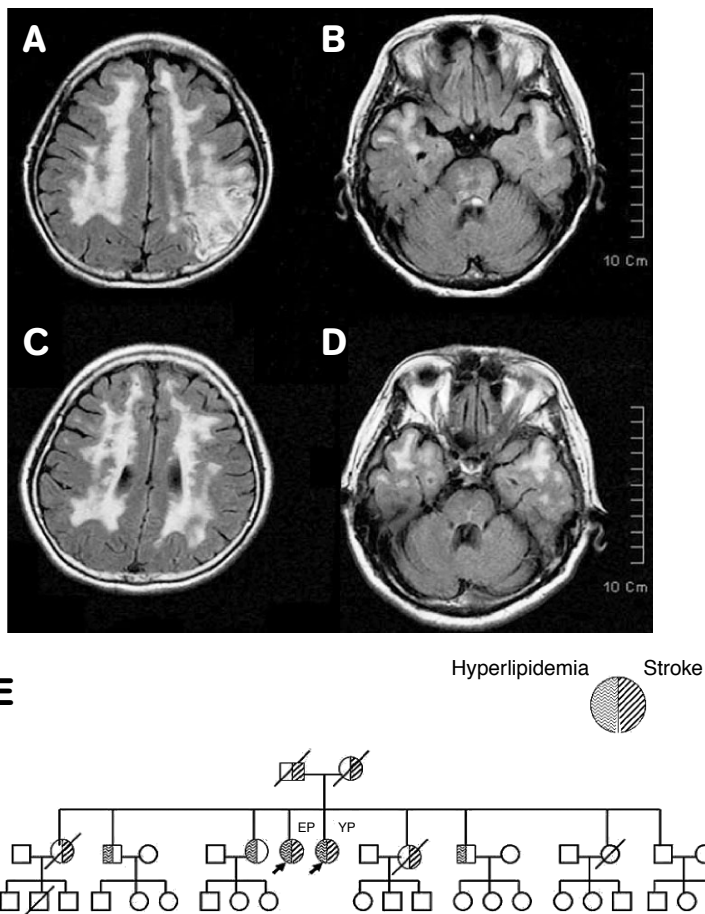


Figure. (A) and (B) showing fluid-attenuated inversion recovery (FLAIR) MRI scans without contrast showing bright signal lesions in the left parietal lobe and extensive confluent hyperintensities in periventricular areas, including bilateral anterior temporal lobes; (C) and (D) showing similar findings in the bilateral anterior temporal lobes and periventricular areas; (E) showing the pedigree of the family, age of the first symptomatic stroke: 49 for II-1, 63 for II-4 and 44 for II-6.

Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL)

is an adult-onset autosomal dominant vascular disease characterized by recurrent strokes, vascular dementia,

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migraine with aura and psychiatric disturbances⁽¹⁾. We here describe a family of CASASIL and demonstrate the magnetic resonance imaging (MRI) of these two patients.

EP is a 63 year-old lady and has experienced at least two cerebral ischemic strokes since October 2007. Her clinical symptoms during this admission included mental deterioration, right hemiparesis, right sensory deficit and pseudobulbar palsy. Her cognitive abilities screening instrument (CASI)-2.0 score was 64/100, with deficits on the items of mental manipulation, attention, and verbal fluency. Her brain MRI showed hyperintensities in the left parietal lobe, bilateral basal ganglia, thalami, brainstem and periventricular area on T2-weighted images, and extensive confluent periventricular T2-fluid-attenuated inversion recovery (FLAIR)-hyperintense lesions, predominantly in the bilateral anterior temporal lobes (Figs. A and B). YP (EP's younger sister) is 62 year-old and had mild psychomotor slowing. Her CASI-2.0 score was 93/100. The brain MRI showed confluent T2-FLAIR-hyperintense lesions involving periventricular white matter, including bilateral anterior temporal areas, and multiple bright signal lesions at bilateral basal ganglia, bilateral thalami and brainstem on T2-weighted images (Figs. C and D). Both patients had hyperlipidemia, but no hypertension, diabetes mellitus, nor migraine. There was neither abnormality in electrocardiography, echocardiography, prothrombotic state including activities of protein C, protein S, and

antithrombin III, venereal disease research laboratory (VDRL), nor antiphospholipid and anticardiolipin antibodies. The family pedigree is shown in Fig. E. Recurrent strokes were found in six members of two generations. EP and YP were proved to have a mutation in Exon 3 of the NOTCH3 gene.

A diagnosis of CADASIL can be made by molecular genetic testing of the NOTCH3 gene, or characteristic granular osmiophilic deposits in vascular smooth muscle cells of skin biopsy⁽²⁾. The involvement of the anterior temporal pole on brain MRI, however, has a sensitivity of 89% and a specificity of 86% for the diagnosis of CADASIL^(1,3). Hence, CADASIL should be highly suspected in patients with recurrent strokes and the aforementioned MRI findings.

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