Neuroimaging Highlight

Editors: W.Y. Hu and M. Hudon

Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL)

Can. J. Neurol. Sci. 1999; 26: 311-312

A patient presented for neurologic evaluation at age 44 following a retro-orbital headache associated with a left homonymous hemianopia that later resolved. He reported migraine with aura headaches, occasionally accompanied by transient aphasia or hemihypesthesia, that progressed in frequency to become a significant disability. His mother and brother both suffered from migraines, and his mother developed a progressive subcortical vascular dementia diagnosed at age 62. Cerebral

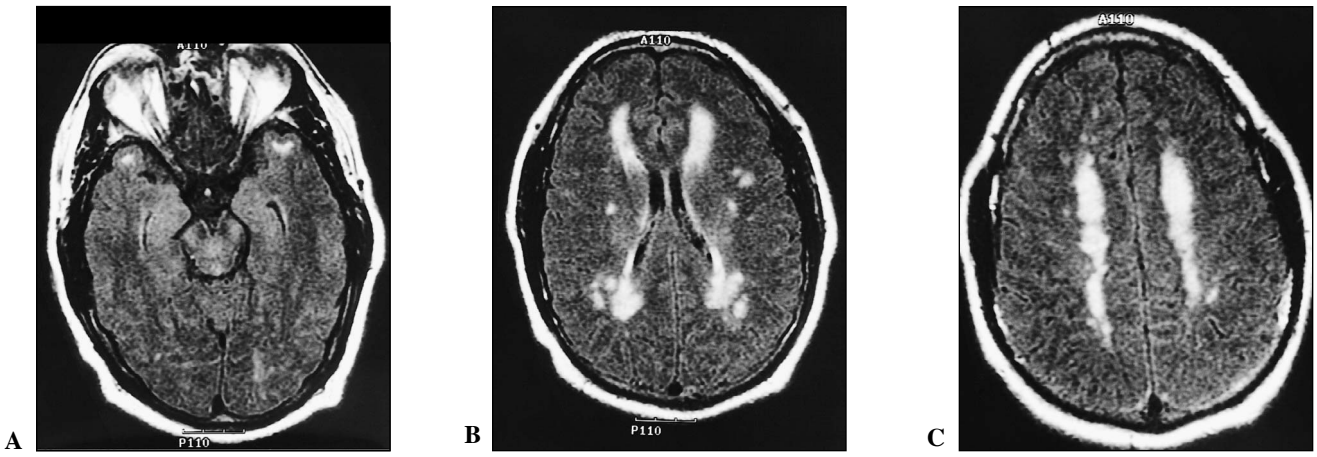

Figure 1. *A-C, Axial FLAIR images in the symptomatic index case reveal abnormal patchy and confluent signal hyperintensities in the subcortical white matter of both temporal lobes (A), periventricular and subcortical white matter (B), and centrum semiovale (C).*

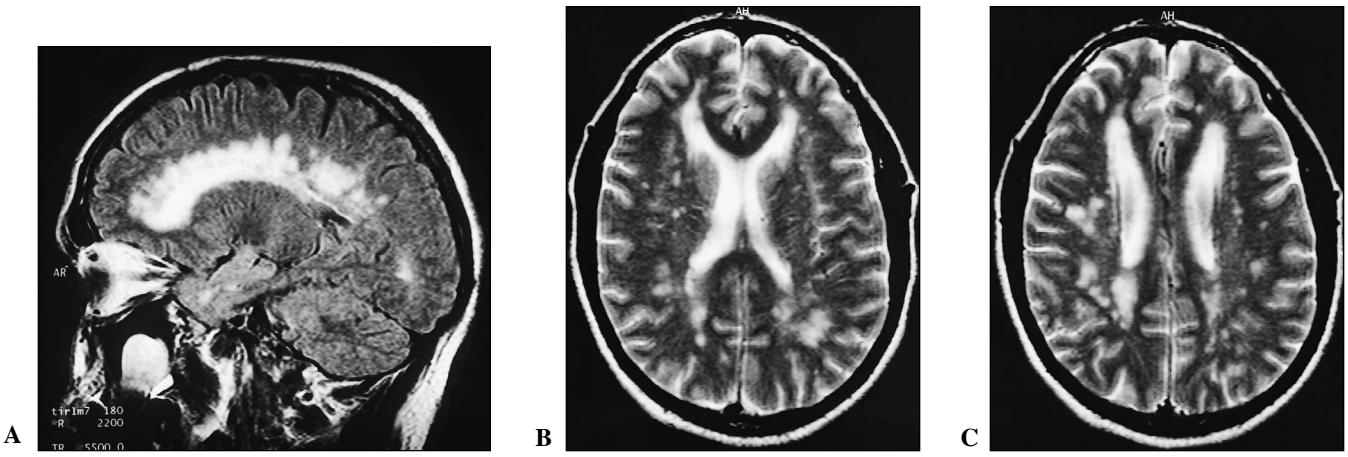

Figure 2. *A-C, Sagittal FLAIR (A) and axial T2 (B and C) images in an asymptomatic male sibling reveal marked confluent signal hyperintensities in the periventicular white matter (A), and more patchy signal hyperintensities in the subcortical and deep white matter (B and C).*

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autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) was suspected and genetic analysis revealed strong linkage to the reported CADASIL locus on chromosome 19q12 in multiple family members.

CADASIL is a dominantly inherited small-artery disease of the brain that causes recurrent subcortical strokes in midadulthood, migraine with aura, mood disorders and eventual dementia. A major feature of this disease is prominent signal abnormalities on MRI with T2-weighted hyperintense lesions in the subcortical white-matter and basal ganglia in all clinically affected subjects (Figure 1) and in asymptomatic carriers (Figure 2) of the mutated gene. Clinical and MRI investigations of family members are important to assist in making the diagnosis, which can then be confirmed by genetic linkage analysis.

Submitted by

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