CADASIL presenting with a movement disorder: A clinical study of a Chilean kindred

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Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) is a hereditary vascular disease that usually begins with migraine, followed by repeated strokes and progressive dementia. We describe an unusual clinical presentation of this condition in members of a Chilean family with an established NOTCH3 mutation. We report early clinical, neuropsychological, transcranial ultrasound, magnetic resonance imaging (MRI), cerebral blood flow, and skin biopsy findings on these patients. Of the patients, 2 presented with facial dystonia, 1 of whom had abnormal single photon emission computed tomography and transcranial ultrasound studies after normal brain MRI scans. Our report emphasizes that CADASIL must be considered in the study of patients with secondary dystonia.