CADASIL: Hereditary vascular dementia. Description of an apparently sporadic clinical case CADASIL: Una forma de demencia vascular hereditaria. Presentación de un caso clínico aparentemente esporádico

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We describe a patient with CADASIL (Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy), the first of four patients diagnosed in our service since 1998, correlating clinical, imaging, and muscular pathology (on electronic microscopy) data. The patient is a woman with no risk factors for cerebrovascular pathology who suffered two transient ischemic attacks at the age of 29. Seven years later, she developed symptoms of depression manifested as psychomotor withdrawal and progressive Parkinsonism that did not respond to pharmacological treatment. An MRI brain scan showed extensive bilateral alterations in the white matter in the frontal and occipital regions, hyperintense in T2 and hypointense in T1-weighted images, without gadolinium enhancement. They had no mass effect, were associated with leukoaraiosis, and were compatible with the diagnosis of CADASIL. Muscle biopsy showed an arteriopathy with CADASIL-type osmiophilic granular deposits in the bl