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INTERESTING MEDICAL IMAGE

CADASIL - Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy

Ramachandiran Nandhagopal

اعتلال الشرايين الدماغية الصبغية الجسدية السائدة مع احتشاءات تحت القشرة واعتلال القشرة والمنطقة البيضاء

راماجاندران ناناجوبال

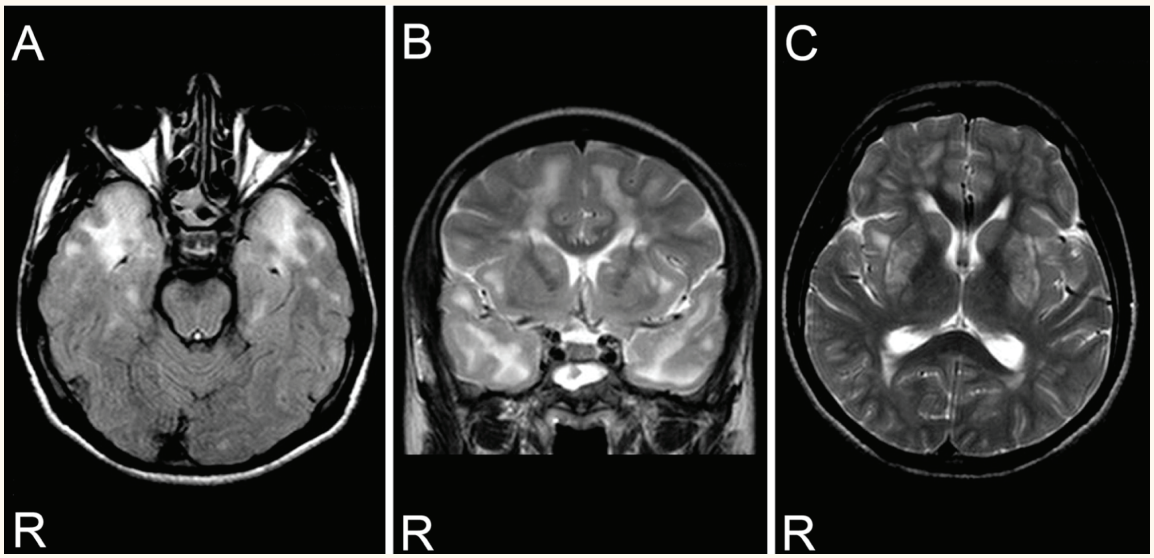

Figure 1: Magnetic resonance imaging (MRI) scan of brain demonstrating increased signal in A) FLAIR image of the white matter in the anterior temporal region, B) frontal region, and C) T2 weighted image of the external capsule and basal ganglia.

A 22 year-old man from South India presented with migraine with aura and depression. Neurologically, he demonstrated cognitive impairment, pseudobulbar affect and bilateral pyramidal signs. The magnetic resonance imaging (MRI) scan of his brain [Figure 1] showed increased FLAIR and T2 signals in the bilateral anterior temporal and frontal white matter, basal ganglia and external capsule. The clinical features and the MRI finding of bilateral white matter lesions in the anterior temporal pole and external capsule, in the absence of hypertension and optic nerve and spinal cord lesions, were diagnostic of cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL).[1-3]

Department of Medicine, Sultan Qaboos University Hospital, Muscat, Oman; Former affiliation: Department of Neurology, Sri Venkateswara Institute of Medical Sciences, Tirupati, Andhra Pradesh, India.

**Correspondning Author email: rnandagopal@yahoo.com*

CADASIL is a rare inherited neurological disorder that clinically manifests with migraine with aura, depression, stroke-like episodes before the age of 60 years, cognitive impairment and behavioral disturbances.[1-4] The characteristic lesions, which can be seen on an MRI scan, involve white matter of temporal pole and external capsule[1-3] and the lesion load is often highest in the frontal white matter.[1-3] Although a positive family history might provide diagnostic support, it is not required in making the diagnosis.[4] CADASIL is associated with *notch 3* gene mutation and *de novo* mutation accounts for the sporadic cases;[4] however, genetic analysis was not available in our centre in India where the patient was seen. Brain MRI scanning aids in the diagnosis of CADASIL in young subjects with a phenotypic manifestation of migraine with aura, depression, stroke and cognitive disturbance, but lacking conventional vascular risk factors such as hypertension, diabetes mellitus, hyperlipidaemia, obesity, smoking and alcohol consumption. The characteristic lesion distribution differentiates these cases from acquired white matter conditions such as multiple sclerosis.

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