



ORAL PRESENTATION

CADASIL – A case report

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Abstract

Introduction: Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL) is the most prevalent monogenic small vessel disease, caused by a mutation in the NOTCH3 gene, situated in chromosome 19. The mutation is probably responsible for a disturbance in vascular mechanotransduction, reducing flow-induced vasodilatation and increasing vascular myogenic tone. In addition, deposition of granular osmiophilic material occurs in smooth muscle cells. With time these alterations will be responsible for several clinical aspects such as migraine with aura, subcortical ischemic events, encephalopathy and psychiatric disturbances, all of which will aggravate with aging. Life expectancy is shortened and only symptomatic treatment is available to these patients.

Clinical Report: A 34 year-old female patient with migraine without aura and sensory deficit after transient ischemic attack (TIA) underwent vascular and genetic investigations after her mother was confirmed with CADASIL. The genetic test confirmed a c.752>A (p.Cys251Tyr) mutation of the NOTCH3 gene, confirming the diagnosis of CADASIL.

Conclusion: Diagnosis of CADASIL is confirmed by the finding of mutations in the NOTCH3 gene. Suspicion must be high upon the presence of family history of stroke, clinical manifestations suggestive of vascular disease and/or suggestive imaging. It is important to think about CADASIL as a differential diagnosis since it is a misdiagnosed disease where confirmation may be obtained long after the development of clinical manifestations. In this case report, the patient already had a pure sensory TIA as well as migraine without aura diagnosed years before she was sent to genetic counselling, where CADASIL was confirmed.

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