



POSTER

CADASIL—an underestimated stroke's cause

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Abstract

Background: Stroke is one of the major causes of death and morbidity. Besides the common sporadic forms related to age, hypertension and atherosclerosis, a minority has a genetic cause, being the most common Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy—CADASIL, caused by mutations in NOTCH3 gene. The diagnose, done in 2-5 in each 100.000, is largely underestimated.

Objectives: This study aims to elucidate the clinical entity CADASIL, highlighting the warning signs that should motivate a more in-depth study and a screening of it.

Methods: Review studies found in Pubmed, between 2013 and 2018 under the keyword “CADASIL” with free full access to the article were considered, 7 were selected.

Results: CADASIL leads primary to migraines followed by premature onset of small vessel ischemic disease, resulting

in vascular dementia, depression, behavioural changes, progressive cognitive impairment and acute encephalopathy. There's no generally accepted diagnostic criteria, but a CADASIL score has been proposed by Pescini et al. that may help confirming the clinical suspicion. Magnetic resonance imaging appearance is characteristic with white matter hyperintensities in the anterior temporal lobe and lacunar infarcts which are an important predictor of cognitive impairment. Definitive diagnosis is given by genetic testing or a skin biopsy. An effective treatment is not available, but the control of vascular risk factors is an important part of CADASIL management.

Conclusions: CADASIL is an underdiagnosed condition that leads to premature and severe morbidity, with characteristic clinical and imagiological findings that should alert the physician to such diagnose, in order to identify individuals at risk and their relatives.

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