Original Paper

Cerebrovascular Diseases

Cerebrovasc Dis 2011;31:77–82
DOI: 10.1159/000321339

Received: February 25, 2010
Accepted: September 1, 2010
Published online: November 5, 2010

Retinal Nerve Fiber Layer Thinning in CADASIL: An Optical Coherence Tomography and MRI Study

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Key Words
CADASIL · Retinal nerve fiber layer thickness · Optical coherence tomography · Brain atrophy · Retinal vessels

Abstract
Background and Purpose: Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) is considered a genetic form of small-vessel disease causing subcortical dementia. A relevant role of axonal injury was recently proposed to explain disability and cognitive decline in this disease. The retinal nerve fiber layer (RNFL) is the only part of the brain where unmyelinated axons can be visualized and quantified in vivo. Their assessment may be an easily reproducible marker of neurodegenerative processes. The aim of this study was to investigate axonal degeneration in CADASIL by measuring RNFL thickness and correlating it with MRI measures of global and regional cerebral atrophy. ***Methods:*** RNFL thickness was measured using optical coherence tomography in 17 CADASIL patients. Average values per quadrant (temporal, superior, nasal, inferior) and overall values were compared with those of normal sex- and age-matched subjects. Data of 13 patients were analyzed for correlations with MRI-based global and regional brain volumes normalized for head size. ***Results:*** RNFL thickness was significantly reduced in CADASIL patients with respect to controls ($p < 0.05$). No significant correlations were found between RNFL thinning and brain atrophy. ***Conclusions:*** RNFL thinning suggests that retinal axonal loss occurs in CADASIL, even in the absence of subjective visual deficit.

Introduction

Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) is considered a genetic model of subcortical ischemic vascular dementia with histological and MRI evidence of white matter changes as hallmarks [1]. Recent data, however, suggest that neuroaxonal loss is critically important in small-vessel diseases including CADASIL. Different quantitative MRI indices of brain tissue damage have been used to estimate neurodegenerative processes in diseases predominantly affecting the white matter. Among these, measures of brain atrophy, which is considered a surrogate marker of cerebral tissue loss, have shown to correlate with disability and cognitive decline better than other MRI measures in CADASIL [2]. Al-

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