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CASE REPORT

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Typical pathological changes of CADASIL in the optic nerve

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Abstract Visual impairment due to retinal and optic nerve changes in cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) is more common than previously thought. Deposits of granular osmiophilic material (GOM) have been shown in the wall of retinal arterioles, though retinal infarcts and vascular occlusions have never been reported. Ischaemic optic neuropathy, on the other hand, has been reported in one case of CADASIL but no pathology reports of the optic nerve have been published. Here we report optic nerve morphological findings in the autopsy material of a 41-year-old woman with genetically assessed CADASIL. Longitudinal and transverse sections of optic nerves were examined. Classical histological methods (haematoxylin-eosin and Nissl) were performed. Diffuse pallor of myelin and rarefaction of optic nerve fibres were observed. Classical GOM was evident in the tunica media of vessels in the meninges and white matter. Arteriole lumina were slightly narrowed. In conclusion, the typical pathological changes of CADASIL occur in the optic nerve and may contribute to impairment of visual function in CADASIL.

Key words CADASIL • Optic nerve • Morphology • Immunocytochemical analysis

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Introduction

Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) is a hereditary late onset disorder characterised by transient ischaemic attacks, migraine, strokes and dementia [1]. The pathological hallmark of the disease is deposition of granular osmiophilic material (GOM) in the tunica media of small arteries and arterioles of the brain white matter and meninges [2]. Smooth muscle cells of arterial walls seem to be the pathological target [3]. Although clinical symptoms are restricted to the central nervous system, the pathological process also affects vessels in skin, muscle, peripheral nerves and internal organs [4]. In recent years, impairment of the visual system has been reported in CADASIL patients. In particular, ophthalmoscopic [5], haemodynamic [6] and electrophysiological [7] changes have been documented in the retina and GOM has been observed in retinal vessels. Beside the retina, the optic nerve may be involved, and ischaemic optic neuropathy was recently reported as a first symptom in a CADASIL patient [8].

Here we report optic nerve morphological findings in autopsy material (orbital, intracanalicular and intracranial portions) of a 41-year-old woman with genetically assessed CADASIL.

Case report

The clinical and neuropathological data of this patient have already been reported [9]. Briefly, at 34 years, episodes of migraine began. At 35 years she experienced right paresis which regressed in a few days. At 36 years, speech impairment associated with ataxia and progressive cognitive impairment were noted. At 40 years, her overall neurological condition deteriorated, leading to a comatose state and death at 41 years. Although she did no complain