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Case Report

Familial Retinal Arteriolar Tortuosity with Acute Hippocampal Infarction

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Keywords

Familial retinal arteriolar tortuosity · Hippocampal infarction · Retinal arteriolar tortuosity · Retinal arteries · Retinal vascular disease

Abstract

Purpose: To report a case of familial retinal arteriolar tortuosity with acute hippocampal infarction. **Method:** Single-patient case report. **Results:** A 50-year-old woman presented with blurred vision and was found to have cataract, retinal hemorrhages, and tortuous retinal arterioles in both eyes. Similar findings of tortuous retinal arterioles were observed in her daughter and son. In her past history of 6 years prior to the visit, she had been diagnosed with transient global amnesia after brain magnetic resonance imaging, which showed hippocampal infarction and multiple chronic ischemic lesions in the periventricular and subcortical white matter. **Conclusion:** Familial retinal arteriolar tortuosity is known to affect the retinal vessels only. To our knowledge, this is the first report of ischemic injury to the brain in a patient with familial retinal arteriolar tortuosity.

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Introduction

Familial retinal arteriolar tortuosity (fRAT) is characterized by tortuosity of the second- and third-order retinal arterioles in the macular and peripapillary areas. This disorder is inherited in an autosomal dominant manner and can be complicated by intra- or preretinal

hemorrhages [1]. Moreover, the systemic abnormalities can be observed in cases of retinal arteriolar tortuosity. Most of the previous studies reported arteriolar tortuosity involving the retina only, but several cases presented systemic abnormalities with retinal arteriolar tortuosity [2–6].

Case Report

We report the case of a 50-year-old woman who presented with a 2-month history of blurred vision in her right eye. BCVA (best corrected visual acuity) was 20/30 and 20/20 in her right and left eye, respectively. An examination revealed lens opacity on her right eye, and a fundus examination showed tortuosity of the retinal arterioles and multiple blot hemorrhages in the perifoveal area. Findings from fluorescein angiography showed arteriolar tortuosity and blocked fluorescence from retinal hemorrhage, but vascular filling defects or hyperfluorescent lesions were absent (Fig. 1). The patient's medical history of 6 years prior to her first visit was noteworthy for a diagnosis of transient global amnesia. She underwent magnetic resonance imaging (MRI) including angiography of the brain at the time of the clinical symptoms and demonstrated multiple chronic ischemic lesions in the periventricular and subcortical white matter, as well as a small infarction in the hippocampus (Fig. 2). Her 26-year-old daughter and 24-year-old son had tortuous arterioles at the vascular arcade without hemorrhage (Fig. 3). The patient underwent cataract surgery for blurred vision in the right eye, and her BCVA recovered to 20/20. At the follow-up 2 months postoperatively, the hemorrhage was completely absorbed.

Discussion

In most cases, fRAT is considered to be an isolated retinal disease. However, associated systemic vascular abnormalities in internal carotid aneurysms, nail bed capillaries, the Kiesselbach nasal septum, and the spinal cord vascular mass, as well as telangiectasis of the bulbar conjunctiva have been reported [3–6]. Sears et al. [3] reported a fRAT patient with migraine. Furthermore, an association with a heterozygous missense mutation in the *COL4A1* gene, which encodes type IV collagen in the basal lamina of various ocular structures and vascular basement membranes, was reported in a family with HANAC (hereditary angiopathy with nephropathy, aneurysm, and muscle cramps). This gene mutation impairs collagen secretion and accumulates misfolded proteins within cells [7]. Such evidence suggests that the vascular malformation may not be limited to the retina.

Our case is a fRAT patient with multiple ischemic lesions on brain MRI. She had been diagnosed with transient global amnesia, which is characterized by the transient loss of the ability to input new memories; a thromboembolic type of cerebrovascular disease is the most widely accepted mechanism [8–10].

Although limitations to the resolution of MRI have made investigating tortuous arteriolar changes difficult, her brain MRI demonstrated multiple ischemic lesions, which may be related to an impairment of cellular diffusion or occlusion of small vessels such as abnormal arterioles. The brain MRI findings for our fRAT patient might be a coincidence. However, our patient has no underlying past medical history of risk factors for cerebrovascular disease and systemic diseases such as diabetes, hypertension, and dyslipidemia. Her medical history

and the early onset of multiple small brain infarctions may suggest a possible involvement of small vascular abnormalities.

Statement of Ethics

The patient/next of kin/guardian has consented to the submission of the case report to the journal.

Disclosure Statement

All authors certify that they have no affiliations with or involvement in any organization or entity with any financial interest (such as honoraria, educational grants, or other equity interest) or nonfinancial interest (such as personal or professional relationships, affiliations, knowledge, or beliefs) in the subject matter or materials discussed in this manuscript.

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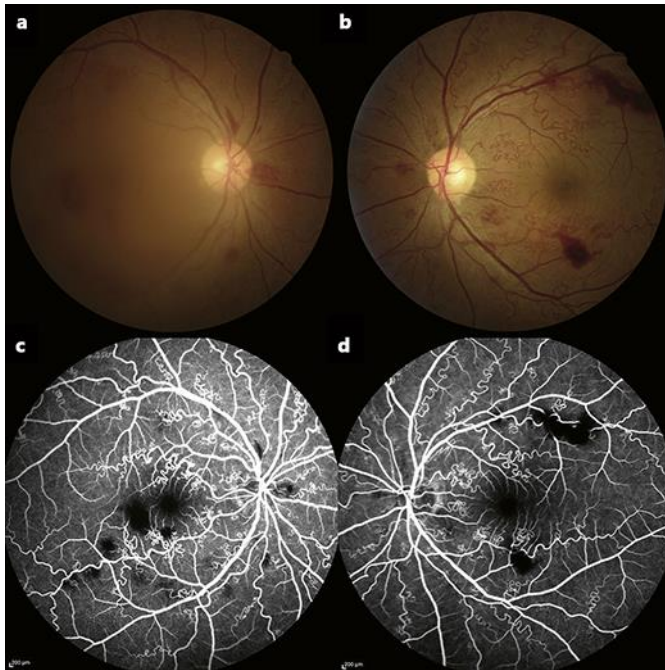


Fig. 1. Fundus photography (**a, b**) and fluorescein angiography (**c, d**) images of the right (**a, c**) and the left (**b, d**) eye. There is a blurrily visible macular area due to lens opacity on the right eye (**a**). Marked tortuosity of the second- and third-order retinal arterioles is present in the macula and peripapillary area, as well as multiple retinal hemorrhages in the perifoveal area of the right (**a**) and the left (**b**) eye. Fluorescein angiography of the right (**c**) and the left (**d**) eye showed tortuous arterioles without leaking and blocked fluorescence due to retinal hemorrhage.

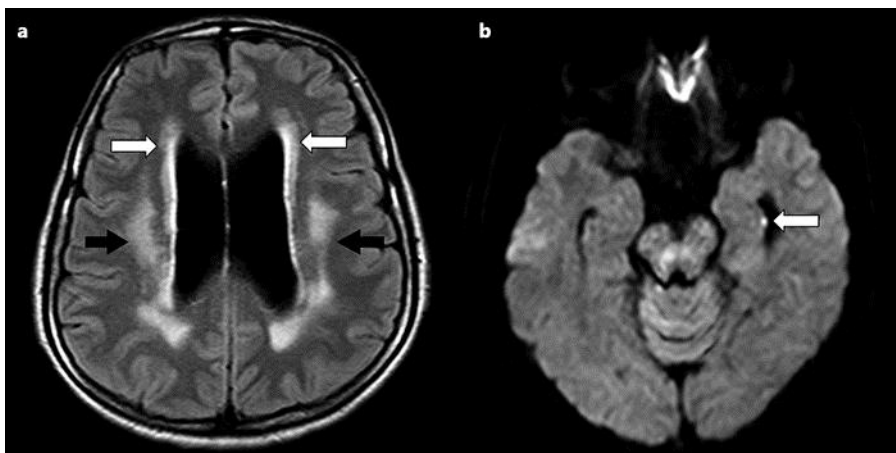


Fig. 2. Brain magnetic resonance images of the patient. **a** Marked multiple chronic ischemic lesions in the periventricular white matter (white arrows) and subcortical deep white matter (black arrows) on T2-weighted imaging. **b** Small, restricted diffusion in the left hippocampus (white arrow) on diffusion-weighted imaging.

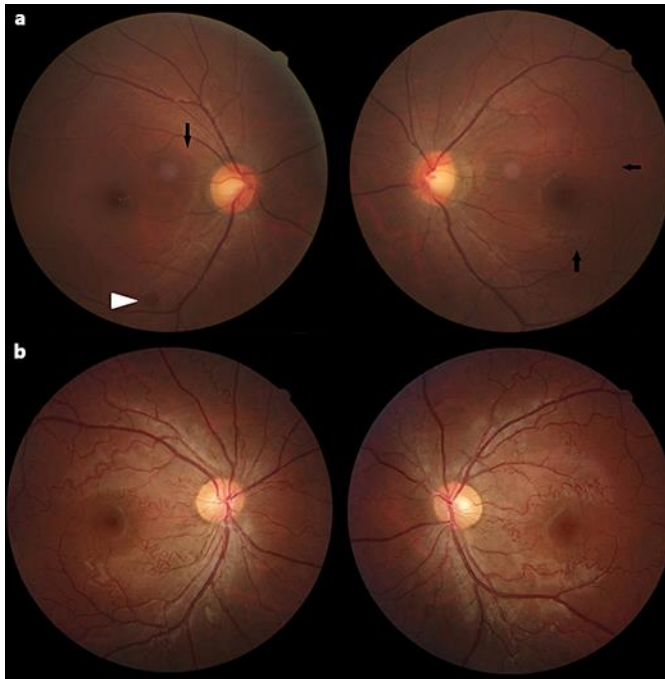


Fig. 3. Fundus photographs of the eyes of the daughter (**a**) and the son (**b**). **a** There is a blot hemorrhage (white arrowhead) on the right eye and mild arteriolar tortuosity (black arrows) on the left eye of the daughter. **b** Marked tortuous arterioles without hemorrhage.