and function in patients with genetically confirmed SCA1. Our data suggest that ATXN1 should be screened in all patients with SCA and decreasing vision. The findings extend the range of ophthalmologic phenotypes and provide important information to assist the management of families in whom SCA1 is suspected.

Veronika Vaclavik, MD
François-Xavier Borruat, MD
Aude Ambresin, MD
Francis L. Munier, MD

Author Affiliations: Department of Ophthalmology, Hôpitaux Universitaires, Geneva (Dr Vaclavik), and Jules-Gonin Eye Hospital, University of Lausanne, Lausanne (Drs Vaclavik, Borruat, Ambresin, and Munier), Switzerland.

Correspondence: Dr Munier, Jules-Gonin Eye Hospital, Avenue de France 15, 1004 Lausanne, Switzerland (francis.munier@fa2.ch).

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Ophthalmic Artery Ischemic Syndrome Associated With Neurofibromatosis and Moyamoya Syndrome

We describe a 12-month-old girl with moyamoya syndrome and neurofibromatosis type 1 who developed profound, unilateral, ophthalmic artery ischemia. The association of moyamoya syndrome with ophthalmic artery ischemia is discussed.

Report of a Case. A 3-month-old girl with neurofibromatosis type 1 was diagnosed as having moyamoya syndrome when she exhibited seizures and, on cerebral angiography, demonstrated right internal carotid artery and middle cerebral artery stenosis with collateral vascularization. Brain magnetic resonance imaging revealed severe right-sided hemiatrophy with laminar necrosis in the right parietal, occipital, and temporal lobes, indicative of a prior ischemic event (Figure 1A). A magnetic resonance angiogram revealed an attenuated right internal carotid artery, severely attenuated right cerebral arteries, and a small right ophthalmic artery with diminished flow (Figure 1B).

At age 4 months, the patient was able to fix and follow in both eyes and exhibited no strabismus. The optic nerve and retinal examination findings were normal. At age 6 months, she underwent a pial synangiosis, a cerebral revascularization procedure in which a donor scalp artery is sutured to the surface of the brain.

Ophthalmic examination at age 12 months revealed a preference for the left eye and a right exotropia. A relative afferent pupillary defect was present in the right eye. Slitlamp examination findings were normal bilaterally.

Dilated funduscopic examination of the right eye revealed a clear vitreous, pale optic nerve, attenuated retinal vessels with abrupt termination of the vessels, and diffuse chorioretinal atrophy nasal to the optic nerve (Figure 2A). Dilated examination of the left eye showed a normal retina, choroid, and optic nerve. Fluorescein angiography of the right eye demonstrated loss of retinal pigment epithelium and atrophy of the choroidal vasculature nasal to the optic nerve, with attenuated retinal vessels (Figure 2B). Findings on fluorescein angiography of the left eye were normal.

Brain magnetic resonance angiography at age 12 months revealed increased stenosis of the right intracranial internal carotid artery and nonvisualization of the right ophthalmic artery.

Comment. Moyamoya syndrome predisposes patients to cerebrovascular ischemia as the result of stenosis of the intracranial portion of the internal carotid arteries and their proximal branches. The development of collateral circulation to compensate for the cerebral ischemia produces an image on cerebral angiography that has been described as a “puff of smoke,” or “moyamoya” in Japanese.

The pathogenesis of the condition is currently unknown, but a polygenic or autosomal dominant transmission with incomplete penetrance has been suggested. The condition has been associated with several disorders, including neurofibromatosis type 1, sickle cell disease, and Down syndrome.

Ophthalmic examination findings associated with moyamoya syndrome include isolated morning glory disc anomaly; a syndrome consisting of morning glory disc anomaly, optic nerve hypoplasia, chorioretinal coloboma, sphenopharyngeal meningoecephalocoele, and midline cranial defects; anterior ischemic optic neuropathy; ocular ischemic syndrome, manifesting with neovascularization of the optic disc, venous dilation and beading, neovascularization of the retina vessels, and vitreous hemorrhage;
Central retinal vein occlusion, and central retinal artery occlusion.

Our patient demonstrated severe optic nerve, retinal, and choroidal ischemia, indicative of an ophthalmic artery occlusion at age 1 year. The patient showed evidence of a prior unilateral stroke but had normal findings on retinal and optic nerve examination at age 4 months. We believe that the ocular ischemia occurred subsequent to this cerebral ischemic event. To our knowledge, this is the first description of a patient with evidence of retinal and choroidal infarction, and consequent necrosis, from ophthalmic artery ischemia associated with moyamoya syndrome and neurofibromatosis type 1. Counseling the patient and family regarding the possibility of development of contralateral disease was performed and close follow-up with neurology and neurosurgery was recommended.

Matthew T. Witmer, MD
Richard Levy, MD
Kaleb Yohay, MD
Szilard Kiss, MD

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Author Affiliations: Departments of Ophthalmology (Drs Witmer, Levy, and Kiss) and Neurology (Dr Yohay), Weill Cornell Medical College, New York, New York.

Correspondence: Dr Witmer, Department of Ophthalmology, Weill Cornell Medical College, 1305 York Ave, 11th Floor, New York, NY 10021 (maw2052@med.cornell.edu).

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