## Ocular Malformations, Moyamoya Disease, and Midline Cranial Defects: A Distinct Syndrome

Sophie J. Bakri, BM, BS, Daniel Siker, MD, Thomas Masaryk, MD, Mark G. Luciano, MD, and Elias I. Traboulsi, MD

PURPOSE: To report a 10-year-old girl with developmental anomalies of both optic disks, a chorioretinal coloboma, sphenopharyngeal meningoencephalocele, and moyamoya disease.

METHODS: A full ophthalmologic examination, cranial magnetic resonance imaging and magnetic resonance angiography, and cerebral angiography were performed.

RESULTS: The patient had a morning glory disk anomaly and microphthalmos of the right eye and optic nerve hypoplasia and retinochoroidal coloboma in the left eye. She had a midfacial cleft and an episode of seizures and a stroke. Magnetic resonance imaging showed a sphenopharyngeal meningoencephalocele. Magnetic resonance angiography and cerebral angiography demonstrated a pattern consistent with moyamoya disease.

CONCLUSIONS: This patient had a distinct syndrome of optic disk, retinochoroidal, and carotid circulation anomalies with midline cranial defects. The recognition and treatment of the vascular abnormalities and cranial defects may prevent complications such as strokes that may occur during or after general anesthesia (Am J Ophthalmol 1999;127:356–357. © 1999 by Elsevier Science Inc. All rights reserved.)

AVITARY OPTIC DISK MALFORMATIONS INCLUDE optic nerve pit, morning glory disk anomaly, and optic nerve dysplasia.<sup>1</sup> They have been associated with systemic abnormalities such as midline facial defects, congenital forebrain abnormalities, agenesis of the corpus callosum, hypertelorism, and renal anomalies, as well as retinal detachment and subretinal hemorrhages.

A 10-year-old girl was evaluated for right esotropia and horizontal pendular nystagmus at age 6 months. She was found to have a morning glory disk anomaly, decreased corneal diameter, and microphthalmos in her right eye. There was optic nerve hypoplasia and an inferior V-shaped retinochoroidal coloboma in her left eye (Figure 1). Visual acuity was RE: 3/200 and LE: 20/40. She had hypertelorism and a mild midfacial cleft with furrowing of the upper lip.

At age 10 years, the patient presented with a 48-hour history of spasmodic movement of the left upper and lower extremities and severe frontal headache. Magnetic resonance imaging disclosed a sphenopharyngeal meningoencephalocele. One month later, she had a stroke. Magnetic resonance angiography and cerebral angiography (Figure 2) showed occlusion of the M1 branch of the right and left middle cerebral arteries and reconstitution of the middle cerebral artery more distally, with delayed filling via tortuous lenticulostriate collaterals and pial collaterals from the posterior cerebral artery. These findings were consistent with a diagnosis of moyamoya disease.

Moyamoya, a Japanese word meaning "cloud of smoke," has been used extensively to refer to a network of small, anastomotic vessels at the base of the brain around the circle of Willis, along with stenosis or segmental occlusion of the terminal parts of both internal carotid arteries. It is a cause of childhood stroke. Most patients present with weakness of a limb and, less commonly, with convulsions, visual disturbances, nystagmus, and headache. Older patients tend to present with subarachnoid hemorrhage.

Four previous cases of an association between optic disk anomalies and carotid circulation anomalies have been reported.<sup>2,3</sup> The atypical infrapapillary V-shaped retinochoroidal coloboma has been associated with basal encephalocele in five patients<sup>4</sup> and with an absent ophthalmic artery in one patient.<sup>2</sup> Our case strengthens the association between morning glory syndrome, other ocular malformations, midline cranial defects, and moyamoya vessels. It also adds evidence that vascular dysgenesis may be an underlying feature of morning glory syndrome.<sup>5</sup> In contrast to all previous cases with unilateral moyamoya disease and morning glory disk anomaly, our patient had bilateral moyamoya vessels and a retinochoroidal coloboma with a hypoplastic optic disk in the fellow eye.

This patient has a distinct syndrome of optic disk, retinochoroidal, and carotid circulation anomalies with midline cranial defects. Vascular and magnetic resonance imaging should be considered in patients with optic disk malformations who present with severe and persistent headaches, seizures, or strokes. Although cerebral angiography is the definitive technique for diagnosing moyamoya disease, less invasive methods such as magnetic resonance angiography or transcranial Doppler may be used. Neurosurgical revascularization techniques are successful in moyamoya disease and avoid the severe impairment from repeated strokes and hemorrhage.

Accepted for publication Aug 31, 1998.

From the Departments of Pediatric Ophthalmology and Strabismus (S.J.B., E.I.T), Anesthesiology (D.S.), Radiology (T.M.), and Pediatric Neurosurgery (M.G.L.), and the Center for Genetic Eye Diseases (S.J.B., E.I.T), The Cleveland Clinic Foundation, Cleveland, Ohio.

Inquiries to Elias Traboulsi, MD, Cleveland Clinic Foundation Eye Institute, 9500 Euclid Ave, Cleveland, OH 44195; fax: (216) 445-8475; e-mail: traboue@cesmtp.ccf.org



FIGURE 1. (Left) Morning glory anomaly of right optic disk with central gliosis and straight vessels emanating from the disk. (Right) Optic nerve hypoplasia with small optic nerve head and inferior V-shaped retinochoroidal coloboma in the left fundus.



FIGURE 2. Left and right carotid arteries and their branches. Note the bilateral stenosis of the middle cerebral artery, which is reconstituted more distally through tortuous lenticulostriate collaterals ("moyamoya vessels"). The "puff of smoke" appearance refers to the network of small anastomotic vessels.

## REFERENCES

- 1. Traboulsi EI, O'Neill JF. The spectrum in the morphology of the so-called "Morning Glory" anomaly of the optic disc. J Pediatr Ophthalmol Strabismus 1988;25:93–98.
- 2. Massaro K, Thorarensen O, Liu GT, et al. Morning glory disc anomalies and Moyamoya vessels. Arch Ophthalmol 1998; 116:253–254.
- 3. Hanson MR, Price RL, Rothner AD, Tomsak RL. Develop-

mental anomalies of the optic disc and carotid circulation: a new association. J Clin Neuroophthalmol 1985;5:3–8.

- Brodsky MC, Hoyt WF, Hoyt CS, Miller NR, Lam BL. Atypical retinochoroidal coloboma in patients with dysplastic optic discs and transsphenoidal encephalocele. Arch Ophthalmol 1995;113:624–628.
- Brodsky MC. Congenital optic disc anomalies. Surv Ophthalmol 1994;39:89–112.