Glaucma Associated With Phakomatoses

A review of glaucoma associated with this group of congenital disorders.

BY DOUGLAS J. RHEE, MD

The term phakomatosis roughly means birthmark, and it refers to a group of conditions that are characterized by hamartomas, congenital tumors arising from tissue that is normally found in that area. The hamartomas can be present at birth or manifest later in life. In general, five phakomatoses are associated with glaucoma: Sturge-Weber syndrome, von Recklinghausen neurofibromatosis, Von Hippel-Lindau disease, nevus of Ota, and phakomatosis pigmentovascularis. Another phakomatosis is tuberous sclerosis, but it is not typically associated with glaucoma.

STURGE-WEBER SYNDROME

General Aspects

The classic hamartomas associated with Sturge-Weber syndrome are a capillary hemangioma of the skin along the trigeminal distribution (also known as a port-wine stain) and ipsilateral leptomeningeal angiomas. The hamartia are typically unilateral but are bilateral in up to 25% of cases.1 Central nervous system (CNS) involvement can lead to seizure disorders and developmental delay. The classic radiologic findings of CNS involvement are cortical calcifications, which appear as double densities or “railroad tracks.” Sturge-Weber syndrome is sporadic with no inheritance pattern.

Ophthalmic

Glaucma is most likely to occur ipsilateral to where the facial angioma is located, especially if there is involvement of the ophthalmic (VI) and maxillary V2 divisions of the trigeminal nerve. The risk of glaucoma is thought to be greatest if the upper lid is involved; generally, eyelid involvement heralds conjunctival involvement (Figure 1) and a presumed increase in episcleral venous pressure when the onset is later in life.

“An estimated one-third to one-half of patients with Sturge-Weber syndrome will develop glaucoma.”

An estimated one-third to one-half of patients with Sturge-Weber syndrome will develop glaucoma.1 When the onset of glaucoma is in infancy, a primary defect of the trabecular meshwork/anterior chamber angle is the pathophysiologic mechanism. A choroidal hemangioma (Figure 2) can also develop and anecdotally is associated with a higher risk of choroidal hemorrhage with incisional surgery. Choroidal and expulsive hemorrhages are thought to occur more frequently in patients with Sturge-Weber syndrome even if there is no choroidal angioma.

Treatment

The infantile form of glaucoma usually requires, and is responsive to, angle surgery (eg, goniotomy or trabeculotomy). For juvenile onset, where elevated episcleral venous pressure is the primary mechanism of the glaucoma, medical treatment generally works poorly. In the latter setting, the author prefers to place a glaucoma drainage device. Trabeculectomy can also be considered, but it carries an increased risk of expulsive and suprachoroidal hemorrhage. Some surgeons prefer to place scleral windows prophylactically imme-

30 GLAUCOMA TODAY JULY/AUGUST 2013
Immediately prior to filtration surgery in case of choroidal hemorrhage.

**VON RECKLINGHAUSEN NEUROFIBROMATOSIS**

**General Aspects**

The classic lesions are (1) café-au-lait spots that are well circumscribed, flat, and hyperpigmented and (2) neurofibromas that are soft, flesh-colored pedunculated masses derived from Schwann cells. CNS involvement is uncommon. Two subsets of von Recklinghausen neurofibromatosis are recognized, both of an autosomal dominant inheritance pattern. The gene is near the centromere on chromosome 17.

**Ophthalmic**

Neurofibromas may involve the eyelids, conjunctiva, iris, ciliary body, and choroid. Hamartomas of iris melanocytes can form cell-defined, clear to yellow or brown, dome-shaped, gelatinous elevations on the iris (also known as *Lisch nodules*). These lesions are generally bilateral. Much as in Sturge-Weber syndrome, the glaucoma is more likely to occur if the eyelids have neurofibromas. The mechanism of glaucoma includes infiltration of the angle with neurofibromatous tissue, angle closure caused by nodular thickening of the ciliary body and choroid, or failure of the anterior chamber to develop.

**Treatment**

Surgical treatment has limited success.¹

**NEVUS OF OTA**

**General Aspects**

The hamartoma of oculodermal melanocytosis (ie, nevus of Ota) is a large accumulation of melanocytes in ocular tissues, especially the episclera (Figure 3). The skin in the distribution of choroidal neovascularization and, occasionally, the mucosa of the nose and mouth can be involved. The condition is unilateral, but bilateral cases occur very rarely. Malignant degeneration is more likely in persons of European descent.¹

**Ophthalmic**

The IOP is elevated in approximately 10% of cases.¹ The involved eye typically has unusually heavy pigmentation of the trabecular meshwork due to the presence of abnormal melanocytes.

**Treatment**

Treatment is the same as typically used for open-angle glaucoma.

---

Douglas J. Rhee, MD, is an associate professor, Harvard Medical School, Massachusetts Eye & Ear Infirmary, Boston. Dr. Rhee may be reached at (617) 573-3670; dougrhee@aol.com.