A Case of Glaucoma Associated with Sturge-Weber Syndrome and Nevus of Ota

Hyo Lee, MD, Seung Sik Choi, MD,* Sung Soo Kim, MD,** Young Jae Hong, MD

The Institute of Vision Research, Department of Ophthalmology, Yonsei University College of Medicine, Seoul,
*Department of Ophthalmology, Pochun CHA University College of Medicine, Pundang CHA hospital, Sungnam,
**Department of Ophthalmology, Ilsan hospital, Koyang, Korea

The Sturge-Weber syndrome consists of a unilateral port-wine hemangioma of the skin along the trigeminal distribution and is accompanied by an ipsilateral leptomeningeal angiomata. Glaucoma is present in approximately half of the cases.

The Nevus of Ota is a melanocytic pigmentary disorder, most commonly involving the area innervated by the trigeminal nerve. Elevated intraocular pressure, with or without glaucomatous damage, is observed in 10% of the cases. We report the first case of glaucoma associated with Sturge-Weber syndrome and Nevus of Ota in Korea.

Key words: glaucoma, nevus of Ota, Sturge-Weber syndrome

INTRODUCTION

The most common vascular lesion associated with the Sturge-Weber syndrome is a port-wine stain or nevus flammeus, which is a vascular malformation that is present at birth. It is composed of ectatic capillaries to venules-sized blood vessels in the dermis. Unlike capillary hemangiomas, the port-wine stain does not resolve over time, and there may be associated blood vessel abnormalities involving the eye on the same side of the face. These may subsequently lead to the development of glaucoma. A leptomeningeal angiomata on the ipsilateral surface of the cerebral hemisphere is the other major component of the Sturge-Weber syndrome. This malformation is responsible for the development of seizures and mental retardation.

The Nevus of Ota is characterized by deep dermal pigmentation, which is usually unilateral. Its area of distribution is to the first and second divisions and occasionally to the mandibular divisions of the trigeminal nerve. Most patients have concomitant hyperpigmentation of the globe, which may involve the sclera, conjunctiva, cornea, iris, and fundus. The iridocorneal angle may be extremely hyperpigmented without an elevation in intraocular pressure.

The association of Sturge-Weber syndrome with the Nevus of Ota is an infrequently reported phenomenon and there are only six previously described cases in the literature. We report the first case of glaucoma associated with Sturge-Weber syndrome and Nevus of Ota in Korea.
CASE REPORT

A 7-year-old boy was referred to the Ophthalmology department of Yonsei University on July 23, 1999, with a glaucoma associated Sturge-Weber syndrome. He had no familial history of neurocutaneous disease. A Nevus flammeus on both portions of the face was present from birth (Fig. 1).

In the initial examination, his corrected visual acuity was 20/50 in both eyes, and the intraocular pressure was 29 and 30 mmHg in the right and left eye, respectively using a Goldmann applanation tonometer. He was bening currently treated with Timolol, Dorzolamide, and Latanoprost at the time. The horizontal corneal diameter was 14mm and the vertical corneal diameter was 13mm in both eyes. A brown pigment deposition in the epithelium of the right inferior cornea was noted (Fig. 2). The left cornea and the anterior chambers of both eyes were clear. Characteristic bluish pigmentation was noted in the conjunctiva and episclera of both eyes (Fig. 3). Brown pigmentation was found at the anterior capsule of lens (Fig. 4). Both fundi were normal and fluorescein angiographies did not reveal a choroidal hemangioma. Gonioscopy showed an open angle on both side but there was heavy hyperpigmentation of the trabecular meshwork in both eyes (Fig. 5). Both optic discs showed a deep cupping with a cup/disc ratio of 0.9(Fig. 6). Visual fields of both eyes showed glaucomatous field defects(Fig. 7). Magnetic resonance imaging which had been previously performed at the other ophthalmology department confirmed that the cortical venous dilatation was due to a venous drainage problem in the left
hemisphere (Fig. 8). It did not show brain atrophy and calcification and no mental defects in the patient were observed. General physical examination revealed an atrophy of the right hand and arm (Fig. 9). Because the intraocular pressure was not controlled by medications, trabeculectomy was performed on the right eye first. We also performed a posterior sclerotomy before entering the right eye during filtration surgery to allow adequate drainage of any choroidal effusion that might occur intraoperatively. Choroidal effusion and expulsive hemorrhage were not noted intraoperatively. One week later, trabeculectomy was performed without a posterior sclerotomy on the left eye (Fig. 10). Melanocyte proliferation which is observed in patients with the Nevus of Ota was found in the scleral specimen that was obtained during the surgery (Fig. 11). At one month postoperative, the intraocular pressure had increased. Timolol was applied to both eyes. Seven months after trabeculectomy, he maintained an intraocular pressure of 14 and 13 mmHg in the right and left eye, respectively using a Goldmann applanation tonometer, and Timolol and Latanoprost in both eyes.

**DISCUSSION**

Sturge described a hemiparetic epileptic girl with a facial angiomia and ipsilateral congenital glaucoma and hypothesized that a cerebral angiomia had caused the neurologic defect. In 1887, Kalischer confirmed the association between facial and intracranial angiomias at autopsy. Weber and Dimitri independently demonstrated radiologic evidence of intracranial calcification. The disease has little familial tendency and no gender or racial predisposition. The Nevus of Ota was first described by Halbe in 1869, before Ota's definitive description in 1939. The Nevus of Ota occurs most commonly in Asians and Black persons and has a strong predilection to occur in females. The coexistence of cutaneous hemangioma and pigmentary nevus

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**Fig. 5.** Heavy hyperpigmentation of the trabecular meshwork

**Fig. 6.** Both optic disc reveal a deep cupping with a cup/disc ratio of 0.9
was termed phakomatosis pigmentovascularis in 1947.\(^6\)

The Sturge-Weber syndrome consists of unilateral port-wine hemangioma of the skin along the trigeminal distribution associated with an ipsilateral leptomeningeal angioma.\(^7\)\(^9\) Glaucoma is seen in approximately half of the cases. The distribution of the port-wine stain is of clinical importance in determining whether an affected child has an increased likelihood of having an eye or central nervous system involvement. Seizures, reflecting the presence of the leptomeningeal angioma, are most likely to be
present if the port-wine stain involves the V₁ area for the branches of the fifth cranial nerve and other areas of the face. Those patients who develop glaucoma in association with a port-wine stain have at least the V₂ area of the branches of the fifth cranial nerve involved. With increasing age, the port-wine stains have a tendency to become darker red or often purplish, and they may become nodular in regions of the hemangioma. The central nervous system vascular malformation is often confined to the pial vessels in the occipitoparietal area. The slow flow of blood through the malformed leptomeningeal vessels leads to a progressive hypoxic injury, encephalomalacia, subsequent atrophy, and calcification in the cerebral cortex. These changes may result in seizure, intellectual impairment, and, less often, contralateral hemiparesis.¹⁰ Seizures are the most common neurologic disturbance, and they develop most commonly during the first year of life. The seizures are predominately of the focal motor type or are generalized and begin on the contralateral side of the body. The most frequent ocular vascular anomaly associated with this syndrome is a hemangioma of the choroid. The conjunctiva frequently have dilated vessels. The most likely cause of the elevated intraocular pressure seems to be a combination of developmental angle anomalies and elevated episcleral venous pressure, which may result in alterations of the meshwork similar to those found with aging. Medical therapy may suffice to control the glaucoma associated with the Sturge-Weber syndrome type that occurs in later life, while the infantile form usually requires surgical intervention. Filtering surgery in these patients is commonly associated with intraoperative choroidal effusion and occasionally with expulsive hemorrhage. Bellows et al suggest performing a posterior sclerotomy before entering the eye when undertaking filtering surgery to allow adequate drainage of any choroidal effusion that might occur intraoperatively.¹¹ Ali and coworkers avoided the occurrence of a choroidal effusion in their cases by using a releasable suture and tightly suturing the scleral flap.¹²

The Nevus of Ota is a melanocytic pigmented disorder, which most commonly involves the area innervated by the trigeminal nerve. The melanocytosis may also affect the oral and nasal mucosa, external auditory canal, tympanic membrane, orbital tissue, meninges, and the brain.¹³ Although the der-
mal hyperpigmentation is usually congenital, it may develop or become noticeable only later in life. Hidano et al reported two peaks of onset, one during the first year of life and the second in the second decade.\textsuperscript{3} Oculodermal melanocytosis has been reported to develop following trauma and sunburn.\textsuperscript{3,4} The reported prevalence of ocular hyperpigmentation in patients with dermal involvement has ranged from 22% to 61.3%.\textsuperscript{15-16} In patients with extensive dermal lesions, ocular involvement is as high as 76.6%.\textsuperscript{3} Elevated intraocular pressure, with or without glaucomatous damage, was seen in 10% of the patients.\textsuperscript{4} The involved eye typically has unusually heavy pigmentation of the trabecular meshwork, and histopathologic studies have revealed melanocytes in the meshwork.\textsuperscript{17-18} Medical management in this, as with other forms of open-angle glaucoma, should be tried first. If this fails, laser trabeculoplasty may be effective, although filtering surgery will most likely be required.

Recent reports suggest that the embryopathogenesis of the skin, leptomeningeal, and choroid and brain lesions of the Sturge-Weber syndrome can be explained by a malformation of the primordial vascular plexus.\textsuperscript{19} In contrast, the hyperpigmentation of the skin, episclera and uvea in Nevus of Ota is thought to be the result of a maldevelopment and abnormality in the migration of neural crest cells, particularly the melanocytes.\textsuperscript{4}

There is evidence for developmental abnormalities of vasomotor nerve cells and melanocytes that originate in the embryonic neural crest in considering phakomatosis pigmentovascularis. Recupero et al have postulated which the association of pigmen
tary and vascular nevi is caused by functional disorders of vasomotor nerve cells and abnormal melanocytes, which originate in the embryonic neural crest.\textsuperscript{20} However, the etiology of phakomatosis pigmentovascularis is unknown. Further investigation into the molecular biology of neural crest development and angiogenesis of the central nervous system is warranted to determine the etiology of these coexisting neuro-oculo-cutaneous disorders.

REFERENCES


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