Case Report

Early Onset and Rapid Progression of Glaucoma in a Neonate with Sturge-Weber Syndrome

Suthida Chaithirayanon MD*, Sumalee Boonyaleephan MD**,
Arucha Treesirichod MD*, Vorasith Siripornpanich MD***

** Department of Pediatrics, Faculty of Medicine, Srinakharinwirot University, Nakhon Nayok, Thailand*
*** Department of Ophthalmology, Faculty of Medicine, Srinakharinwirot University, Nakhon Nayok, Thailand*
**** Research Center for Neuroscience, Institute of Molecular Biosciences, Mahidol University, Nakhonpathom, Thailand*

Sturge-Weber syndrome (SWS) is an uncommon neurocutaneous syndrome usually presenting with a triad of cutaneous, neurological, and ophthalmological symptoms. The cutaneous lesion can be observed at birth in most cases while the symptoms of the nervous and ocular systems involvement usually appear later in life. The most common ocular manifestation in SWS is glaucoma, which can occur in the early-life period. The authors reported a case of SWS in which the symptoms of glaucoma rapidly developed within two weeks following an ophthalmologic evaluation that was initially negative at the age of one week.

Keywords: *Sturge-Weber syndrome, Glaucoma, Port-wine stain, Ocular symptoms, Neonate*

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Sturge-Weber syndrome (SWS) is a congenital syndrome that manifests with cutaneous, neurological, and ophthalmological symptoms. The cutaneous manifestation in SWS is a facial port wine stain (PWS) which usually appears at birth as an erythematous patch on the ophthalmic division (V1) of trigeminal nerve (CN V). However, only a few cases of patients exhibiting a PWS are diagnosed as SWS. The appearance of a PWS in conjunction with neurological as well as the presence of ocular symptoms are required in the diagnosis of SWS. Bilateral PWSs, extension of a PWS to other territories of CN V and PWS with involvement of the upper eyelid are the risk factors for SWS[1]. Although the PWS is the most concerning problem among parents of affected children, the ocular and neurological symptoms are more serious complications. No factors can be used to predict the onset or progression of the ocular or neurological consequences of this disease. Here, the authors present a case with rapidly developing symptoms of glaucoma, the most common ocular complication in SWS, at the age of three weeks with a previously normal eye examination during the perinatal period.

Correspondence to:
Siripornpanich V, Research Center for Neuroscience, Institute of Molecular Biosciences, Mahidol University, Nakhonpathom 73170, Thailand.
Phone: 0-2441-9003-7 ext. 1311, 1206
E-mail: drvorasith@gmail.com

Case Report

A three week-old Thai female neonate presented with erythematous patch on the left side of her face with distribution to the ophthalmic (V1) and the maxillary divisions (V2) of the fifth cranial nerve at birth. The child's birth and mother's maternal histories were unremarkable. Initial neurological evaluation was normal and no additional abnormalities were detected during the child's early neonatal period. She was sent to an ophthalmologist at the fifth day following her birth for glaucoma screening. In the initial examination, intraocular pressures (IOP) were 20 mmHg in both eyes by use of a Tonopen. Her corneal diameters were 10 mm in both eyes. There were no buphthalmos or evidence of cloudy corneas in both eyes. Both optic discs showed a cup/disc ratio of 0.3. However, her mother had noticed an enlargement of the child's cornea in her left eye beginning at the age of two weeks. Ophthalmologic evaluation at three weeks revealed a mildly clouded cornea, an enlarged corneal diameter, and a raised IOP in the left eye. Thus, the glaucoma was diagnosed. She was referred to the university hospital in Bangkok for surgery. The surgeon's first attempt was to perform a nasal approach 120-degree goniotomy when she was only two months old. Unfortunately, her IOP in the left eye remained high. Thus, a second goniotomy was performed at 120 degrees approaching at the temporal side of her left eye three months later. Finally, the IOP in her left

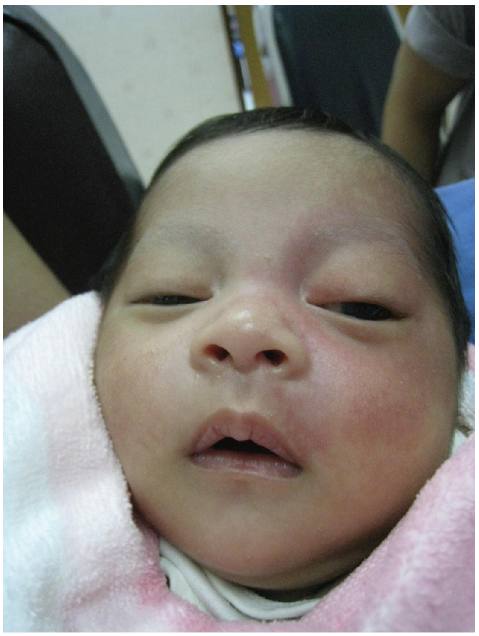

Fig. 1 Clinical photograph showed facial port-wine stain over the left side of patient's face. This picture was taken at the first day of life.

eye was 16 mmHg with the use of only one anti-glaucoma medication. At her one-year follow-up, the IOP in her left eye remained under control.

During the period of ophthalmologic treatment, she also underwent neurological investigations in an attempt to find additional data for diagnosing SWS. Although the result of her electroencephalography (EEG) was normal, CT scans of the brain demonstrated hyperdense cerebral gyri at the right frontal cortex and bilateral intravascular enhancements with predominance on the left side. At around the age of five months, she developed focal tonic-clonic seizures of her left hand and received Phenobarbital for controlling her seizure symptoms. Hence, the triad of SWS symptoms was complete.

Discussion

SWS is a sporadic neurocutaneous syndrome that commonly manifests as a triad of symptoms consisting of a facial port-wine lesion, venous angiomatosis of the leptomeningeal, and concurrent ocular abnormalities. The facial PWS is caused by arteriovenous malformations in the skin. It occasionally involves the conjunctival, episcleral, retinal, and choroidal structures. Although the facial vascular abnormality is usually unilateral, between 10% and 30% are bilateral[2,3].

The most common ocular complication in SWS is glaucoma, which can present at any age but usually manifests with bimodal distribution: early- and late-onset forms. Glaucoma in SWS predominately shows its effects unilaterally, on the same side as the PWS, especially in cases exhibiting upper-lid involvement[4]. Another ocular manifestation in SWS is a diffuse choroidal vascular malformation that leads to visual loss resulting from progressive retinal damage.

Childhood glaucoma is an uncommon disease in the general pediatric population. The incidence of glaucoma in people under the age of 20 is only 2.29 cases per 100,000 people[5]. However, the risk to developing glaucoma is increased in patients with SWS, particularly in cases with PWS in both the V1 and V2 dermatomes[6]. Glaucoma has been reported in 30 to 71% of SWS patients[7-9] and approximately 60% have congenital glaucoma with buphthalmos, and advanced optic nerve cupping. Forty percent of patients develop glaucoma later in childhood or in adulthood[7,8,10]. The pathogenesis of glaucoma in SWS is still unknown. Histopathological studies in the congenital glaucoma group have shown ranges from; a poorly developed scleral spur, a thickened uveal meshwork, or an anteriorly displaced iris root. Late onset glaucoma patients often have normal-appearing angles where the iris meets the cornea.

Weiss et al[11] has proposed two mechanisms for the development of glaucoma in SWS. In congenital glaucoma, he hypothesized that raised episcleral venous pressure from episcleral angiomas combined with abnormal angle development lead to the development of congenital glaucoma. In late onset cases with normal-appearing angles, he thought that the cause of glaucoma was due to the elevation of the episcleral venous pressures[11-13]. In the present case, the symptoms of glaucoma could be detected at about the age of three weeks. Interestingly, the first ophthalmologic evaluation during the early neonatal period was normal, but our patient presented her ocular symptoms two weeks later with rapid clouding and enlargement of the cornea of the left eye.

Although congenital glaucoma is more common in patients with SWS, the rapid progression of the ocular symptoms as presented in our case is unusual. One study has published that mutation in CYP1B1 gene, encoded for an enzyme in the cytochrome P450 superfamily, is associated with congenital glaucoma. Mutation in this gene indirectly affects aqueous outflow by disrupting the development

and turnover of the extracellular matrix of the trabecular meshwork or can directly contribute to abnormal elevations of the IOP by disrupting the metabolism of ciliary body-derived mediators in these processes[14]. Therefore, the authors analyzed the CYP1B1 gene mutations in this case and the result was negative, similar to a recent study[15] that has reported no CYP1B1 mutations in unilateral buphthalmos with glaucoma.

The management of glaucoma in patients with SWS consists primarily as surgical intervention and has been difficult. Goniotomy and trabeculotomy should be attempted first in congenital glaucoma with buphthalmos for controlling IOP. Unfortunately, these procedures are often ineffective. Most patients may need additional surgeries or medications to control their IOP. Thus, early detection and prompt treatment are the key factors for a good visual prognosis in patients with glaucoma.

Furthermore, advances in the management of cutaneous lesions in SWS with the use of laser treatments has become a topic of debate with concern to the risk of developing ocular hypertension and glaucoma following laser treatments of the PWS. However, the latest study did not find any significant association between laser treatments and development of glaucoma in SWS patients[16].

In conclusion, as we have learned from the present case, the authors emphasize the importance of frequent ophthalmologic evaluations in children with PWS as well as the monitoring of the symptoms of glaucoma that can be rapidly developing and progressing in the early life period.

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Potential conflicts of interest

None.

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ภาวะต้อหินที่เกิดขึ้นในช่วงแรกของชีวิตและเปลี่ยนแปลงอย่างรวดเร็วในทารกที่เป็นโรค SWS

สุธิดา ชัยธีระยานนท์, สุมาลี บุณยะลีพรรณ, อรุชา ตรีศิริโชติ, วรสิทธิ์ ศิริพรพาณิชย์

โรค Sturge-Weber syndrome (SWS) เป็นโรคในกลุ่ม neurocutaneous ที่พบไม่บ่อย มักมีอาการหลัก 3 ด้าน ประกอบด้วย อาการทางผิวหนัง อาการทางระบบประสาท และอาการทางตา อาการทางผิวหนังสามารถพบได้ตั้งแต่แรกเกิด ในขณะที่อาการของระบบประสาทและอาการทางตามักจะปรากฏเมื่อโตขึ้น อาการทางตาที่พบได้บ่อยที่สุดของโรค SWS คือ ภาวะต้อหิน ซึ่งสามารถเกิดขึ้นได้ในตั้งแต่ช่วงแรกของชีวิต คณะผู้นิพนธ์รายงานผู้ป่วยโรค SWS ที่อาการของภาวะต้อหินเกิดขึ้นอย่างรวดเร็วภายในสองสัปดาห์ หลังการตรวจตาครั้งแรกให้ผลลบเมื่ออายุ 1 สัปดาห์