

Case Report

A rare case of bilateral ocular manifestations of Sturge-Weber syndrome

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ABSTRACT

Sturge-Weber syndrome (SWS) is a group of phakomatoses characterized by hamartomas involving brain, skin and eyes. A 36 years old female presented with pain and diminution of vision in both eyes since last 1 year. On examination she has bluish sclera in B/L eyes with characteristic facial port-wine stain. Best corrected visual acuity (BCVA) in R/E- 6/18, N6 and in L/E- 6/6, N6. Intra-ocular pressure (IOP) in R/E – 27 mm Hg and in L/E – 18 mmHg. On gonioscopy – open angle B/E. On direct ophthalmoscopy and slit-lamp examination with 90 D lens, cup-disc ratio in R/E – 0.9 and in L/E – 0.7 with thinning of neuro-retinal rim in B/E. In optical coherence tomography (OCT), retinal nerve fibre layer (RNFL) and optic nerve head (ONH) it was confirmed. Other systemic examination – within normal limit. Initially, she had been on conservative treatment. As intraocular pressure remained high after several weeks of treatment, trabeculectomy in R/E done under local anaesthesia. No post-operative complications were seen. After 7 days of follow-up IOP – within normal limit, vision in R/E unchanged and had been frequently followed-up.

Keywords: Sturge-Weber syndrome, Phakomatoses, Hamartomas, Intraocular pressure, Gonioscopy, Trabeculectomy

INTRODUCTION

Sturge-Weber syndrome (SWS) belongs to a group of disorder known as phakomatoses. These are usually hamartomas which are congenital tumours that arise from the tissue that normally found in the involved site. Other disorders included in phakomatoses are neurofibromatosis, Klippel-Trenaunay syndrome, tuberous sclerosis, and von Hippel-Lindau syndrome.¹ It is also known as encephalotrigeminal angiomas. It includes three classical manifestations like leptomeningeal hemangioma, facial angiomas or nevus flammeus (also called port-wine stain [PWS]), and pathological ocular changes.²⁻⁵ SWS has no hereditary pattern like other phakomatosis and there is a report of involvement of somatic mutation in the G protein subunit alpha Q (GNAQ) gene.⁶ The incidence of SWS is ~1:50,000 infants, with no significant difference between males and females.⁷

The embryologic basis of SWS is found to be maldevelopment of the cell precursors in neural crest cell during the first trimester that can lead to the pathologic manifestations observed in the central nervous system, skin, and eyes.³ Diagnosis is easily performed when the classical clinical signs of SWS are present, consisting of unilateral facial PWS along the first branch of the trigeminal nerve, hemiatrophy, progressive seizures, contralateral hemiparesis, mental retardation, hemianopia, and ipsilateral glaucoma.⁸ However for confirmatory diagnosis of SWS there should be at least two manifestations of classical triad (leptomeningeal angioma, PWS, and eye abnormalities). Clinical manifestations of SWS may vary in different cases with or without ocular involvement.⁹

According to the clinical manifestation, SWS is classified into four types: presence of brain and facial angioma, with or without glaucoma; PWS without central nervous system

(CNS) involvement, with or without glaucoma; isolated brain angioma without glaucoma; and type 1 associated with systemic manifestations.¹⁰ Ocular manifestations of SWS comprise vascular abnormalities of eyelid, orbit, conjunctiva, episclera, ciliary body, retina and choroid. Approximately 50% of SWS patients show pathologic ocular changes, usually ipsilateral to the PWS.³ Iris heterochromia with hyperpigmentation of the affected side may be noted. Glaucoma is the main challenging ocular abnormality of SWS. Glaucoma usually has unilateral presentation and mostly diagnosed during infancy, but there is occurrence of late onset glaucoma also during adolescent or adulthood in few case reports.

We herein report a case of bilateral ocular manifestation of SWS with adult onset glaucoma.

CASE REPORT

A 36 years old female presented at outpatient department (OPD) of Assam Medical College and Hospital, Dibrugarh with chief complaints of pain in both eyes since 1 year. It was insidious in onset, mild in nature and non-progressive. She also complained of diminution of vision in both eyes for last 1 year. Diminution of vision was gradually progressive in nature more in the right eye than in left eye. There was no H/O redness and watering of eyes. There was also no history of trauma or any previous ocular surgical procedure in the eyes.

For her diminution of vision she first visited a private clinic where she came to know her disease condition and she was prescribed spectacles and medications for the same and was asked for regular check-up.

There was no similar history in the past or such history in her family as stated by her. But she said that the hyperpigmentation on her face was present since her childhood but no other symptoms were present. There was no other systemic symptoms as she said. She belongs to a lower middle class family according to Kuppuswamy scale living with her husband.

She was brought to eye OPD of Assam Medical College and Hospital as she complained she was unable to see clearly with spectacles also.

Examination revealed a visual acuity of 6/36 in the right eye and 6/18 in the left eye on Snellen's chart. Best corrected visual acuity (BCVA) in right eye 6/18, N6 and in the left eye 6/6, N6. On examination, findings were bluish discolouration of sclera in B/E as shown in Figure 1 and dilated episcleral veins in R/E which was visible with normal torch light examination in Figure 2. Intra-ocular pressure (IOP) in the R/E 27 mmHg and in L/E 18 mmHg on Goldmann applanation tonometer. Gonioscopic examination revealed open angle in both eyes. Fundus examination with direct ophthalmoscope and on 90 D slit-lamp examination revealed cup:disc ratio in the R/E approximately 0.9 as shown in Figure 3 and in

L/E 0.6. Thinning of neuro-retinal rim in both eyes was observed. No other abnormality was detected. Optical coherence tomography (OCT) of optic nerve head (ONH) and retinal nerve fiber layer (RNFL) of both eyes confirmed cupping and RNFL thinning which is more in right eye than left eye as shown in Figure 4. Humphrey visual field (HVF) 30-2 analysis in R/E showed double arcuate scotoma and than in left eye no visual field defect was detected showing below in Figure 5. HVF 10-2 was proposed for R/E.



Figure 1: Port-wine stain in B/E.

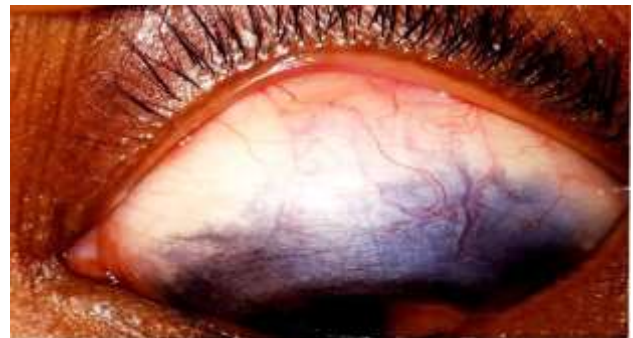


Figure 2: Dilated episcleral vein.



Figure 3: Fundus picture of R/E.

She was initially given medical treatment in her first visit at the private clinic. It included- eye drop travoprost- 1 drop once daily in R/E at 9 pm; eye drop brimonidine tartrate and timolol maleate – 1 drop 12 hourly in R/E, and brinzolamide ophthalmic suspension (IP 1.0% w/v) – 1 drop 12 hourly in R/E.

When she was taken to eye OPD of Assam Medical College and Hospital then looking at her severity and resistance to medical treatment surgery trabeculectomy was proposed and at a later date trabeculectomy was done in the right eye under local anaesthesia.

On follow-up after day 7, IOP in R/E was 18 on applanation tonometry and vision was unchanged. No other complications were noted. Patient has been asked to follow-up at regular interval.

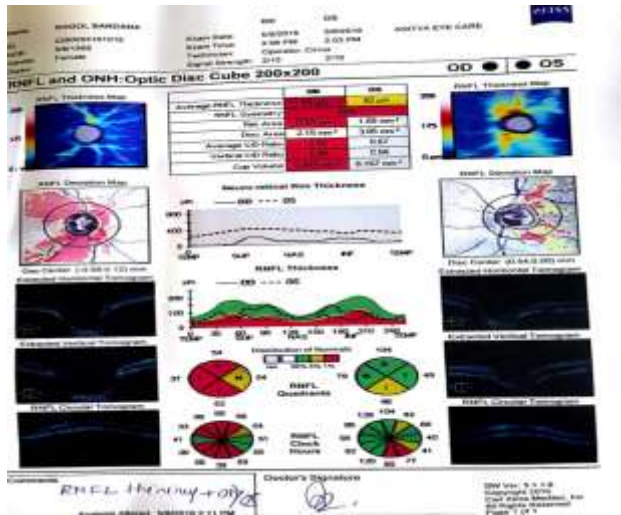


Figure 4: OCT RNFL and ONH – RNFL thinning present R/E > L/E.

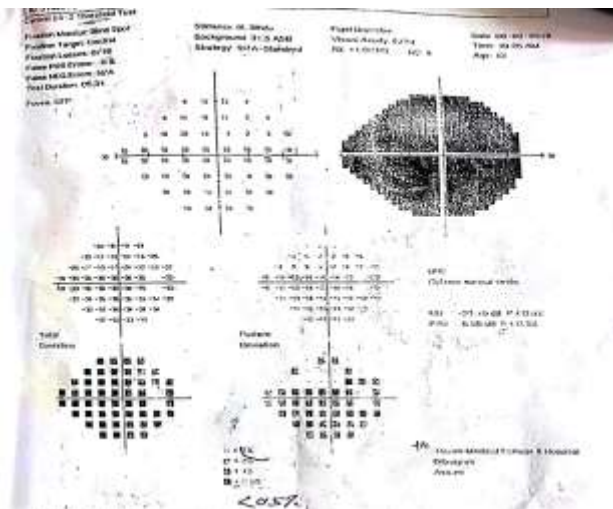


Figure 5: HVF 30-2 of R/E.

DISCUSSION

Approximately 50% of SWS patients show ocular changes. Ocular changes are usually ipsilateral to the PWS and the eyelid, anterior chamber, cornea, choroid, and retina are usually involved.³ To diagnose a case of SWS it should follow at least two criteria of the classical triad (leptomenigeal angioma, PWS, and eye abnormalities). However, in different cases clinical findings of SWS may

be different with variable neural signs and symptoms and there may be absence of ocular involvement.⁹

PWS is usually present on the forehead and upper eyelid along with maxillary and ophthalmic distribution of trigeminal nerve. It is usually unilateral and present at birth but the lesion can extend both sides of the face and extremities like in our case. With increasing age it becomes darker red or deep purple with associated vascular ectasia. Waelchli et al showed that the PWS distribution may follow the embryonic vasculature distribution of the face, rather than along the distribution of trigeminal nerve.¹¹ Other neurological manifestations like gyriform calcification, neuroangiomatosis and astrogliosis in brain are not present in our case.

There is a report of development of glaucoma in 30–70% of SWS cases.^{12,13} Glaucoma shows a bimodal peak of age development. One is early-onset (congenital) form affecting ~60% of patients and another peak is a later-onset form affecting children and adolescence (40% of cases).¹² In our case, glaucoma is adult onset and it is bilateral. The posterior segment of the eye is also involved with hemangiomas of the choroid (20%–70% of cases).¹⁴ In our case no other abnormal fundus findings are present except glaucomatous optic disc and RNFL changes.

There are many new hypotheses and factors on the pathogenic mechanisms that can lead to the development of glaucoma.

The formulated theories of development of glaucoma in SWS include the following.

A mechanical mechanism related to congenital malformation of the anterior chamber angle leading to increased resistance to aqueous humor outflow. In this case, the iris may also not have the flat anterior insertion resembling congenital form of glaucoma.¹³ It is usually associated with buphthalmos, anisometria and amblyopia.

A rise in episcleral venous pressure (EVP) because of arteriovenous shunts mechanism into the episcleral hemangioma. This theory is based on the observation of a normal angle structure, blood within Schlemm's canal, and more severe glaucoma and it is mainly responsible for the adult onset glaucoma.¹⁵⁻¹⁷

Hypersecretion of fluid either by the ciliary body or the choroidal hemangioma.¹⁸

A variation from normal hemodynamics of the anterior chamber angle and episclera because of premature aging of the trabecular meshwork–Schlemm's canal complex in later-onset glaucoma.¹⁹

In our case we can be attributed that the cause of glaucoma is because of elevated episcleral venous pressure as postulated by Weiss as the main cause of late onset glaucoma.¹⁶ Another study by Phelps in 16 eyes with SWS

it was observed that the cause of glaucoma was mainly because of elevated episcleral venous pressure and episcleral hemangioma.¹⁵ Tannous et al in their study reported that in late-onset glaucoma, increase in episcleral venous pressure may be due to progressive hypertrophy and dilatation of the episcleral veins.²⁰ Maruyama et al reported a case of SWS with acute glaucoma along with posterior scleritis, ciliochoroidal effusion, edema of the ciliary body and anterior rotation of the ciliary body.²¹

Facial PWSs can be successfully treated by laser. Laser should be done superficially to avoid potential complications like decrease in brain venous outflow through PWS vessels, leading to deterioration of cerebral veins, dilatation of choroidal vessels, exudative retinal detachment, and rise in IOP. Therefore, deep photocoagulation and debulking surgery should be avoided in the treatment of PWS.²²

Glaucoma associated with SWS is more difficult to manage than other forms of glaucoma with a lower success rate and increased risk of complications. The main goal of management of glaucoma is to reduce IOP and to prevent further damage of optic nerve and visual field. Because of rare nature of the disease, it is more difficult to manage with topical antiglaucoma drugs mainly in congenital form however it is the first line of therapy in late onset form.¹⁷

Ong et al showed in their study of 14 patients with SWS that latanoprost eye drops, as adjunctive therapy, were effective in controlling glaucoma in 50% of cases during 1 year of follow-up.²³ Latanoprost acts by increasing uveoscleral outflow, thereby bypassing the obstacle to the passage of aqueous humor due to the increase in EVP. In our case travoprost eye drop was used which is another prostaglandin analogue. Along with travoprost eye drop brimonidine tartrate and timolol maleate and brinzolamide ophthalmic suspension (IP 1.0% w/v) were also used in our case but there was no improvement seen with topical therapy. The use of other antiglaucoma drugs like beta-blockers and carbonic anhydrase inhibitors is found to be effective in some SWS patients with absence of buphthalmos in few case reports.²⁴

In case of early onset glaucoma with anomalous anterior chamber angle surgical treatment like goniotomy or trabeculotomy is usually preferred. If topical treatment does not produce adequate response then in late onset glaucoma also surgical intervention is required and glaucoma filtration surgery trabeculectomy remains the procedure of choice as it bypasses the episcleral venous system. Outcomes may be improved with antimetabolites but there is a long term risk of blebitis and bleb related infection. In addition, in SWS patients trabeculectomy is associated with higher risk of choroidal effusion due to higher pressure difference. In our case also reduction in IOP was seen after trabeculectomy during follow-up period and no surgical complications were noted. However vision was not improved in our case as there was already

double arcuate scotoma defect in the visual field as shown in HFA analysis of right eye.

There are some alternative surgical interventions like nonpenetrating sclerectomy, drainage valve implants, and ciliodestructive procedures in adults in case of intractable glaucoma.^{25,26} In SWS, the Ahmed-type valve was shown to induce long-term decrease of IOP by improving aqueous humor outflow.²⁷

Moreover, in SWS, the surgical success rate is the lowest among secondary glaucomas, since surgical failure, uncontrolled IOP, and low vision outcomes have been frequently reported.¹⁸ In spite of availability medical and surgical approaches to treat both forms of glaucoma, the development of this ocular complication still represents the worst prognostic factor for vision loss in SWS patients.

CONCLUSION

SWS is a life time disease though the patients may be asymptomatic throughout their childhood. Glaucoma is the most common ocular complication. Indeed, because of its early development and weak response to standard medical therapy surgery is frequently required to obtain long-term control of IOP in order to avoid visual function loss. Moreover, in SWS, the surgical success rate is the lowest among secondary glaucomas. Due to the rare nature of the disease and SWS-related ocular conditions, much of the data presented here is from small studies and case report. With better data, the need for multidisciplinary care and surveillance could be fulfilled.

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