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Original Research Paper

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STURGE-WEBER SYNDROME ASSOCIATED WITH CENTRAL SEROUS RETINOPATHY: A RARE CASE SCENARIO

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ABSTRACT **Aim:** To report a rare case with Sturge-Weber Syndrome associated with central serous retinopathy. SWS is a rare sporadic genetic disease and diagnosis is primarily done by evaluating history, the presence of port-wine stain, and characteristic features on brain radio imaging. As no definitive treatment is available yet, patients are being treated by medical and surgical interventions for symptoms as well as for associated complications. **Material And Methods:** 18-year-old male presented with blurred vision and was found to have elevated intraocular pressure (IOP) and glaucomatous disc damage in the left eye. He also displayed capillary malformation (port-wine stain) on the same side of the face. IOP got reduced after prescribing a topical anti-glaucoma drug. This case exhibits a very rare occurrence of Sturge-Weber syndrome affecting the left side of the face associated with ipsilateral central serous retinopathy. **Result:** The patient responded well to the topical beta-blocker in the subsequent follow-up when the IOP was found to be well controlled. Central serous retinopathy also got reduced in 1 month with no changes in BCVA. **Conclusion:** Sturge-Weber syndrome is a rare phacomatosis that may present with central serous retinopathy.

KEYWORDS : Sturge-Weber syndrome (SWS), Intra-ocular pressure (IOP), Best corrected visual acuity (BCVA)

INTRODUCTION

Encephalo-trigeminal angiomatosis is another name for the congenital neuro-ocular-cutaneous condition known as Sturge-Weber syndrome. Facial nevus, ocular abnormalities (choroidal hemangioma, glaucoma), and leptomeningeal angioma are its distinguishing features[1]. an illness that affects 1 in every 50,000 live births and is extremely uncommon.

Clinical examination, history taking, and radio imaging (CT scan and MRI) are the main methods used for diagnosis. On these tests, the typical signs of calcification and leptomeningeal enhancement can be noticed. There is no preference based on ethnicity or gender.

Here, we discuss the case of a male patient, age 18, who complained of impaired vision and whose increased intraocular pressure (IOP) and left eye glaucomatous disc damage was later discovered.

On the same side of his face, he also had capillary malformation (port wine stain). In this case, ipsilateral central serous retinopathy and Sturge-Weber syndrome, both extremely rare occurrences, damage the left side of the face.

Case Report

An 18-year-old male complained of blurred vision in his left eye for 1 month. He also gave a history of redness in his left eye since childhood. His best-corrected visual acuity (BCVA) was 20/20 in the right eye and 20/30 in the left eye. Anterior segment evaluation of both eyes was unremarkable except for a scleral nevus seen in the left eye.

[Fig. 1] Fundus examination revealed a cup-to-disc ratio of 0.3:1 in the right eye and 0.6:1 in the left eye. On applanation, tonometry intraocular pressure (IOP) was found to be 14 and 27 mmHg in the right and left eye.

A four-mirror gonioscopy revealed open angles. The retinal examination revealed central serous retinopathy in the left eye which was an accidental finding. [Fig. 2]

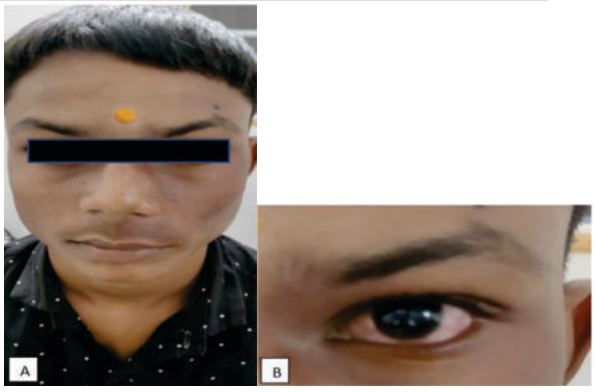

Figure 1: Picture (A) showing a Port wine stain on the left side of the face and (B) Scleral nevus

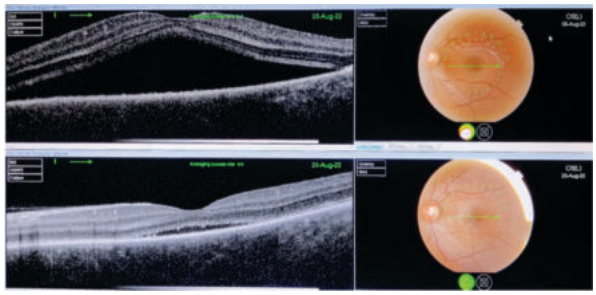

Figure 2: Comparison of one-month-apart OCT pictures showing Central serous retinopathy.

Table 1: Routine blood Investigations

Investigations	Values
Hemoglobin (gm %)	12.5
Total Count (per cumm)	9,300
Differential Count (N/L/E/M %)	54/44/01/01
Platelet Count (per cumm)	3,73,000
Random Blood Sugar (mg/dl)	90
Urea (mg/dl)	25
Creatinine (mg/dl)	0.87

Total Bilirubin (mg/dl)	1.0
Sodium (mmol/L)	140
Potassium (mmol/L)	4.7
Magnesium (mg/dL)	1.8
Ionized Calcium (mmol/L)	1.31
Urine Routine Micro	Normal

Table 2: Radiological Investigations

Investigations	Findings
Ultrasonography of the Abdomen and Pelvis	Normal
Skull X-ray	Normal
CT Head (Plain)	Normal
MRI Brain (Plain+ Contrast)	Normal
Electroencephalogram (EEG)	Normal

A conjunctival scar and erythematous lacy capillary network-like lesions covering the left side of the face were discovered during a physical examination.

The brain's magnetic resonance MRI did not reveal any obvious abnormalities.

The patient was identified as having Sturge-Weber syndrome together with central serous retinopathy based on typical ocular characteristics.

He was started on timolol eye drops 0.5% two times a day in the left eye. The patient responded well to the topical beta-blocker in the subsequent follow-up when the IOP was found to be well controlled.

Central serous retinopathy also got reduced in 1 month with no changes in BCVA.

DISCUSSION

We discuss a rare instance of central serous retinopathy and Sturge-Weber disease. Sturge-Weber is an embryonic developmental defect caused by GNAQ gene mutation, mesodermal, and ectodermal development abnormalities. [2,3]

In addition to neurological issues, Sturge-Weber syndrome is distinguished by a birthmark called a facial nevus or port-wine stain. It typically runs along one or more trigeminal nerve segments unilaterally. Although typically unilateral and visible at birth, the lesion may also affect the extremities and both sides of the face. PWS appears flat and pale pink in neonates. In age, it changes color to a deep purple or a darker red and develops vascular ectasias that are nodular and hypertrophic. Current PWS management involves using a pulsed dye laser.

10–20% of babies with a typical PWS develop leptomeningeal capillary-venous malformation. [4] Affected individuals also exhibit a "tram-line" look on MRIs that may be a leptomeningeal angioma, which is typically linked to a convulsive condition and occasionally to contralateral hemiplegia and mental retardation. In many situations, patients experience neurological symptoms later in childhood or maturity, and the diagnosis could go unnoticed without the proper neuroimaging.

The most prevalent significant eye condition, glaucoma, affects 30–70% of people with Sturge–Weber syndrome[5] and is brought on by abnormalities of the anterior chamber angle or excessive episcleral venous pressure. It is the primary underlying mechanism of late-onset glaucoma. [6] The patient in the current investigation exhibited left-eye glaucomatous cupping.

Complete Sturge-Weber syndrome is defined as having both CNS and facial angiomas, whereas incomplete Sturge-Weber syndrome only has one afflicted area. Almost half of the choroid is often affected by diffuse choroidal hemangiomas, which grow very slowly. occurs ipsilateral to the nevus almost exclusively in patients with Sturge-Weber disease. The hue of the fundus is a deep, diffuse red. Exudative retinal detachment, subsequent retinal cystoid degenerations, and neovascular glaucoma are complications.

When a patient is seen to have a face port-wine birthmark connected to ipsilateral brain or eye involvement, the diagnosis of Sturge-Weber syndrome is commonly suspect. Due to the central serous retinopathy, unilateral port-wine stain, and ipsilateral glaucoma seen in our instance, it is intriguing.

SWS is not specifically treated. Pulsed dye laser photo coagulation for port-wine stains and symptomatic and preventative treatment for glaucoma are among the medical services provided in SWS. Patients with refractory seizures, glaucoma, or a specific illness connected to one of the SWS-associated disorders are more likely to benefit from surgery. Focused cortical resection, hemispherectomy, corpus colostomy, vagal nerve stimulation, and surgical intervention for diffuse choroidal hemangiomas with retinal detachment and glaucoma are surgical procedures for SWS.

CONCLUSION

The most frequent ocular consequence of SWS is glaucoma. Congenital glaucoma is the most common kind, although it can also affect adults and children, necessitating thorough ocular monitoring of SWS patients. Central serous retinopathy may be present in SWS, an uncommon phacomatosis.

Acknowledgment

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Conflict of interest

There is no conflict of interest.

REFERENCES

(1) Baselga E (2004). Sturge-Weber syndrome. Semin Cutan Med Surg;23:87-98.
(2) Moe P, Seay AR (2003). Current pediatric diagnosis and treatment;16:717-92.
(3) Shirley MD, Tang H, Gallione CJ, Baugher JD, Frelin LP, Cohen B, et al. N Engl J Med 2013;368:1971-9.
(4) Comi AM. *Neurologist*. 2011;17(4):179-184
(5) Budnez DL et al. *Ophthalmology*.2000;107(11):2105-2110.
(6) Weiss DI. *Trans Ophthalmol Soc UK*.1973;93(0):477-493.