TO REPORT A CASE OF STURGE WEBER SYNDROME TYPE 2 IN A YOUNG FEMALE

DR. BHUPINDERJEET SINGH[1]
CO-AUTHORS-DR. REENA KUMARI[2], DR. DINESH KR BHAGAT[3], DR. KHUSHDEEP KAUR[1]
[1]POST GRADUATE TRAINEE (FINAL YEAR)-DEPARTMENT OF OPHTHALMOLOGY, [2]PROFESSOR-DEPARTMENT OF OPHTHALMOLOGY,
[3]ASSOCIATE PROFESSOR-DEPARTMENT OF OPHTHALMOLOGY
MATA GUJRI MEMORIAL MEDICAL COLLEGE AND LSK HOSPITAL, KISHANGANJ, BIHAR

INTRODUCTION

Sturge-Weber syndrome (SWS), also referred to as encephalotrigeminal angiomatosis is a rare disorder which is characterized by angiomas that can involve face, eye and brain. It belongs to a group of disorders known as phacomatoses. SWS can present with variable phenotypes in different individuals [1]. The incidence is estimated to be 1 in 50,000-60,000 live births [2]. The GNAQ gene which is present on the long arm of chromosome 9 undergoes somatic mutation which leads to this syndrome [3]. According to Roach scale classification, it is of three types:

•**type I** is characterized by angioma involving both face and brain with or without pre-existing glaucoma;

•**type II** is characterized by angioma involving face only (no CNS involvement) with pre-existing glaucoma;

•**type III** is characterized by leptomeningeal angiomas, no facial angioma with or without the presence of glaucoma.

CASE REPORT

A **18 years old female** presented to Otorhinolaryngology department for **oral ulcers** and was referred to Ophthalmology department. She complained of progressive **loss of vision** in **left eye** since 2 years associated with dark red discoloration on left side of face.
Patient was apparently normal 2 years back when she developed diminution of vision in left eye which was insidious in onset and progressive in nature.
No history of trauma and drug intake. There was no significant past or family history.
On examination by torch -
Port wine stain was noted involving the whole **left** side of **face**, extending to right mandibular region too. There was **lower lip hypertrophy** also (Figure 1).

Investigations performed were-
- Slit lamp examination(anterior segment shows **dilated and tortuous episcleral vessels** (Figure 2).
- Goldmann applanation tonometry-**IOP**-Right eye-12mmhg and **left eye-22mmhg**.
- Gonioscopy shows both eyes have **open angle** in all quadrants with multiple iris processes.
- Brightness(B) scan ultrasonography-both eyes-unremarkable.
- OCT-right eye-unremarkable; **left eye**-shows deep cup, **cup disc ratio-0.9,** bayonetting sign, retinal nerve fibre layer thinning (Figure 3 and 4 respectively).
-HFA 24-2 threshold (Figure 5 and 6).
-MRI-Brain with contrast (Figure 7).

Anterior segment examination

	Right eye	Left eye
Vision unaided	6/6	6/18
Lid	Normal	Port wine stain
Conjunctiva	Clear	Dilated and tortuous episcleral vessels
Cornea	Clear	Clear
Anterior chamber	Normal depth	Normal depth
Iris	Central Heterochromia	Central Heterochromia
Pupil	Round, regular and reactive	Round, regular and reactive
Lens	Clear	Clear
Extraocular movements	Full and painless	Full and painless



Figure 1: shows port wine stain and lower lip hypertrophy.



Figure 2: Left eye- showing dilatated and tortuous episcleral vessels and central heterochromia of iris.

Posterior segment examination

Right eye	Left eye
• Media clear • Disc- Normal size and well defined margin • Cup disc ratio-0.4 • Retinal vessels- normal • Foveal reflex – present	• Media clear • Disc- deep cup • Cup disc ratio-0.9 • Retinal vessels – nasal shifting of vessels, • Bayonetting sign-present • Foveal reflex present

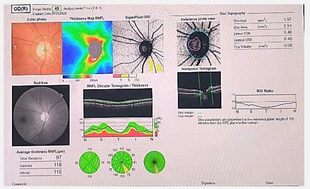


Figure 3: OCT shows optic disc parameters within normal limit.

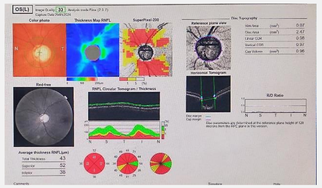


Figure 4: OCT shows deep cup, cup disc ratio-0.9, bayonetting sign, retinal nerve fibre layer thinning.

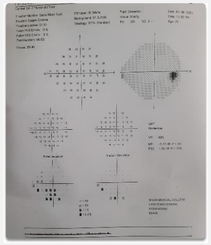


Figure 5: Right eye HFA 24-2-unremarkable

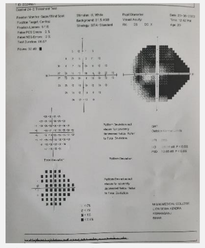


Figure 6.-Left eye HFA 24-2 shows peripheral visual field defect – tunnel vision defect

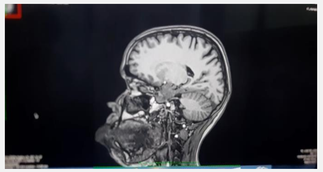


Figure 7: MRI Brain with contrast shows swelling of subcutaneous tissue over facial region with no leptomeningeal involvement.

DIAGNOSIS

On the basis of slit lamp examination, Optical Coherence Tomography (OCT) and Magnetic Resonance Imaging (MRI), diagnosis of Sturge Weber Syndrome type 2 was made, with ocular manifestations of **open angle glaucoma**, dilatated episcleral vessels, **central heterochromia** in left eye and central heterochromia in right eye.

TREATMENT

-Eye drop **Timolol maleate 0.5%** 1 drop BD daily in left eye was started and was followed weekly for a month. IOP measurements were taken which showed a drop by approximately 30% and then she was counselled for 3 monthly follow-up.
-For port wine stain, patient was referred to dermatology department for appropriate treatment.
-For oral ulcers, patient was treated by ENT department with Betadine mouth wash 5cc BD for 5 days, Triamcinolone Acetonide oral paste LA for 10 days.
-Psychiatric care was also established for the patient.

DISCUSSION

SWS is a multisystem disorder that can present with neurological, cutaneous, ocular, and oral manifestations. **Cutaneous manifestation**-Port wine stain is the most common clinical manifestation of SWS [4]. It is typically located over the face but can sometimes be widespread and is present since birth. **Ocular manifestations-**Development of glaucoma in SWS patients shows a bimodal peak :~ 60% of patients can present at birth i.e. an early onset form or congenital form and 40% can present later in life i.e. a later-onset form during childhood and adolescence[5]. Glaucoma in SWS is unilateral and is associated with the presence of an ipsilateral Port wine stain,[6] which can involve both the eyelids (72%) or sometimes only the upper eyelid (21%).-The most common form of glaucoma in SWS patients is open-angle glaucoma leading to progressive visual field loss. Other ocular manifestations can be heterochromia of the iris, dilated and tortuous episcleral vessels and choroidal haemangioma. **Neurological manifestations** can be leptomeningeal angiomatosis which is usually unilateral involving parietal and occipital region. Investigations such as CT and MRI of the brain with gadolinium are most useful in addition to skin and eye examination. Multidisciplinary treatment of SWS is essential for optimal management including expert psychological evaluation and counseling of patients and their families as port wine stain can cause psychological trauma to the patient.

Clinical manifestations of SWS and manifestations seen in our case.	
1 Epilepsy	-
2 Port wine stain	+
3 Leptomeningeal abnormality	-
4 Oral manifestations	+
5 Ocular manifestations	+
6 Mental retardation	-

CONCLUSION

Sturge Weber Syndrome is a rare congenital disorder that occurs due to somatic mutation in GNAQ gene. Early detection of Sturge Weber Syndrome is essential to detect development of complications like glaucoma that can lead to total blindness, choroidal haemangioma and other systemic manifestations.

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