INTERNATIONAL JOURNAL OF HEALTH & MEDICAL RESEARCH

ISSN(print): 2833-213X, ISSN(online): 2833-2148

Volume 05 Issue 04 April 2026

DOI : 10.58806/ijhmr.2026.v5i4n07

Page No. 320-322

Secondary Glaucoma in Patient with Sturge Weber Syndrome at Tertiary Hospital, North East, Nigeria: A Case Report

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ABSTRACT

Background: Sturge-Weber Syndrome (SWS) is a congenital, and sporadic neurocutaneous syndrome occuring as a result of angiomas involving the leptomeninges, and the skin of the face along the ophthalmic and maxillary divisions of the trigeminal nerve. It is characterized by a facial port-wine stain presenting at birth, and glaucoma is a common ocular manifestation.

Method: Patient clinical notes from electronic medical records with a diagnosis of SWS was used and the relevant literature reviewed. Informed consent was obtained from the child's parents, and ethical approval obtained from the Hospital Ethical Committee.

Result: An 8 year old female child presented with history of loss of vision, pain and photophobia in the left eye of three (3) years duration. Parents had noticed dark patch on the left side of her face at birth, and later tearing and gradual enlargement of eyes noticed during one year of life. On examination, she had port-wine stain involving the left side of her face. Left eye examination revealed unaided visual acuity (VA) of hand movement (HM), with tortuous conjunctival and episcleral veins, marked corneal epithelial oedema, and a dilated and sluggish pupil. Further view of the lens and posterior segment was not clear due to a hazy media. Intraocular pressure (IOP) was 10 and 35mmHg in the right and left eye respectively.

Conclusion: sws is a rare condition and glaucoma is a frequent associated clinical manifestation which if treatment is not commenced on time would lead to blindness. All patients with facial discolouration/port-wine stain in the ophthalmic distribution of the trigeminal nerve that present to the eye clinic should have an evalution to rule out glaucoma during all visits and subsequent regular visits advised.

In our clinical setting, treatment may be commenced based on typical clinical symptoms,port-wine stain, and clinically confirmed glaucoma.

KEYWORDS: sturge weber syndrome, congenital, brain, pot-wine stain, glaucoma

INTRODUCTION

Sturge Weber Syndrome is a neurocutaneous syndrome, chaacterized by the association of facial port-wine hemangiomas in the trigeminal nerve distribution area, with a vascular malformation of the brain leptomeningeal hemangioma, with or without glaucoma.[1]

It is a rare condition with an estimated incidence of 1 in 20,000- 50,000 live births.[2] It is said to be associated with a somatic mutation in the GNAQ gene[3] that results in an association of a facial capillary malformation, and/or a vascular malformation of the brain.[4]

The syndrome is classified clinically into: a trisystem/Type I (facial, leptomeningeal angiomas, and glaucoma); bisystem/Type II (facial angiomas alone, may have glaucoma). The condition's hallmark features include a facial port-wine stain, neurological abnormalities such as convulsions and intellectual disability, and glaucoma.[5]

This report presents the rare case of an 8-year-old female child diagnosed with SWS, highlighting the clinical presentation, diagnostic workup, management strategies and the role of multidisciplinary approach in the management of the condition.

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CASE REPORT

An 8 year old female child residing in Gombe town of Gombe State, North-eastern Nigera.

The patient was first seen at the GOPD clinic of the FTH Gombe three (3) years ago (in 2021) with history of loss of vision in the left eye. She was placed on eyedrops (?name) but patient was later lost to follow-up.

She presented again one year ago (April, 2023) at the GOPD clinic with same complaints and was referred to the eye clinic. Patient was first seen at the eye clinic in May, 2023 with complaints of visual loss and whitish discolouration of black part of her left eye. Other clinical details of history and examination was not documented, and a diagnosis of left eye end-stage glaucoma was made and patient was placed on Gutt. Misopt.

Further review by consultant ophthalmologist one (1) month later revealed a history of loss of vision, pain, redness, and photophobia in the index eye. Parents had noticed dark patch on left side of her face at birth, and later tearing and gradual enlargement of eyes was observed during the first one year of her life. There was no prior history of eye trauma. No history of convulsions, and other review of systems and paediatric history was nil of note. There was also no similar history among family members and marriage was non-consanguineous. On general examination: patient had stable vital signs, a non-tender, raised and erythematous (reddish to purple coloured) discolouration of the skin that involved the left side of forehead, half of nose, cheeks and left eyelids along the distribution of the trigeminal nerve.

Ocular examination findings in the right eye were essentially normal. Unaided VA was 6/6 and HM in the right and left eyes respectively. IOP was 10 and 35 mmHg taken at 11.55am. Left eye had mild buphthalmos with tortuous conjunctival and episcleral veins, marked corneal epithelial oedema, normal depth anterior chamber, dilated and sluggish pupil, iris/lens details not clear (due to hazy media), posterior segment details was not clear due to hazy media. Imaging studies (MRI, CT, B-Scan) was deferred (due to family's financial constraint). A diagnosis of Sturge Weber Syndrome (Bisystem: portwine stain + end stage glaucoma) was made.

Patient was placed on dorzolamide + timolol combination and flurbiprofen eyedrops. Patient's parents were also counselled on possible diagnosis, treatment outcome, and genetic nature and implication of disease. Subsequent follow—up visits revealed poor IOP control, and so patient waa counselled for possible LE Trabeculectomy in orderto achieve optimal IOP control in view of family financial contraints and treatment options available within the facilty.[6]

DISCUSSION

SWS is a rare, congenital neurocutaneous disorder characterized by a facial port-wine stain, leptomeningeal angiomatosis, and ocular abnormalities. These abnormalities represent hamartomatous capillary malformations. The Roach scale classifies the cndition clinicallyinto into: Type I (facial, leptomeningeal angiomas, and glaucoma- trisystem); Type II (facial angiomas alone, may have glaucoma- bisystem); Type III (isolated leptomeningeal angiomas, no glaucoma).

A seizure is usually the earliest neurological manifestation of SWS with infantile spasm seen in approximately 90% of patients in the first year of life followed by myoclonic seizures later. Other neurological symptoms are; hemiparesis, recurrent headaches, stroke, and mental retardation.[7] Port wine stain (flaveus naevus) is present at birth, it is typically unilateral and does not change with the age of the patient (in contrast to infantile hemangioma- not present at birth, and involutes later).[8] Glaucoma is ipsilateral to the port-wine stain, other ocular findings are congestion of the conjunctiva, tearing, and globe enlargement. Not all patients with PWS have SWS, and patients with SWS may present with only cerebral symptoms without PWS.[9] In our case report, PWS was noticed at birth and eye symptoms suggestive of early onset glaucoma was observed during her first year of life. There was no history of convulsions however.

Evalution and diagnosis of SWS is based on typical clinical symptoms, facial appearance, ophthalmic examination to rule out a secondary open-angle glaucoma, buphthalmos etc., and magnetic resonance imaging (MRI) and computed tomography (CT scan).[4]

A contrast enhanced T1-weighted MRI will show leptomeningeal vascular malformation, and a Brain- CT which will show cortical and subcortical calcification is the basis of diagnosis. However, radioimaging studies may not be remarkable in patients with Type II SWS (bisystem) who have only facial angiomas and glaucoma (like in our case report). In such cases, diagnosis may rely mainly on clinical evaluation and genetic testing (where available). Other imaging studies such as EEG, magnetic resonance spectroscopy fluorodeoxyglucose-positron emisssion tomography (FDG-PET) may also help in evaluation of patients but are not routinely used.

Treatment is multidisciplinary and mainly supportive as there is no specific treatment for SWS. Interventions range from medical (and sometimes surgical) management for seizuress; special education programmes for patients requiring cognitive support; physiotherapy for hemiparesis; laser therapy for cosmetic management of the port-wine stain (or counselling for possible emotional/psychological trauma caused by the cosmetic blemish); and medical/surgical management for glaucoma. Treatment

for glaucoma is usually with topical antiglaucoma drugs or surgery. The decision for trabeculotomy, goniotomy, trabeculectomy, glaucoma drainage devices etc., will depend on age of onset, severity, and other factors.[10]

Despite the fact that SWS is a rare condition, this case reported is even more rare as majority of the patients with SWS have cerebral/neurological involvement and present with convulsionsin 90% of cases. Radioimaging in this case may not be conclusive on the diagnosis and a high index of suspicion is therfore required in similar patients presenting with secondary glaucoma and port-wine stain. However , other differential diagnosis e.g. Infantile Hemangioma, Von Hippel Lindau disease, Mafucci syndrome, angio-osteodystrophy syndrome, Klippel Trenaumy-Weber syndrome, Rendu-Osler-Weber syndrome, Wyborg Mason, PHACES, Blue Rubber bleb naevus syndrome, should also be entertained and ruled out.

CONCLUSION

SWS is a rare condition but presents a unique set of challenges due to its multisystem involvement as well as its need for expensive and sometimes unavailable radio maging tests. Glaucoma is a frequent associated clinical manifestation which if treatment is not commenced on time would lead to blindness.[2] Therefore, clinicians should have a high index of suspicion for SWS in all patients presenting with port wine stain, and evaluate extensively based on typical clinical symptoms, facial appearance, ophthalmic examination to rule out glaucoma. Also, all patients with SWS should be screened for onset of glaucoma at regular follow-up visits as they may develop glaucoma at any point of their lifetime.[11]

This case also highlighted the importance of a comprehensive, individualized treatment plan for patients with Sturge-Weber Syndrome. Multidisciplinary care involving neurology, ophthalmology, dermatology, and supportive services is essential for managing the complex clinical features of SWS and improving patient outcomes.

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