Sturge-Weber Syndrome in A 23-Year Old Nigerian Male: A Case Report

***Chinawa NE[1], Edoga CE[2]**

ABSTRACT

Sturge-Weber syndrome (SWS, also called encephalofacial or encephalotrigeminal angiomatosis) is a neurocutaneous syndrome, characterized by the association of facial port-wine hemangiomas in the trigeminal nerve distribution area, with a vascular malformation of the brain (leptomeningeal angioma) with or without glaucoma. Herein, we reported Sturge-Weber syndrome in a 23-year-old man, who presented wi-th a right port-wine hemangiomas, ipsilateral iris hyperchromia, suspected choroidalhaemangioma and glaucoma.

Keywords: Vascular malformation, sturge-weber syndrome, encephalafacial

INTRODUCTION

Sturge-Weber syndrome (SWS, also called encephalofacial or encephalotrigeminal angiomatosis) is a neurocutaneous syndrome, characterized by the association of facial port-wine hemangiomas in the trigeminal nerve distribution area, with a vascular malformation of the brain (leptomeningeal angioma) with or without glaucoma[1]. It was first described by Schirmer and later more specifically by Sturge in 1879. It is also known as Sturge-Weber disease, encephalotrigeminal angiomatosis, meningofacial angiomatosis, and Sturge-Weber-Dimitri syndrome[2].

It is a congenital disorder occurring due to dysfunction of embryonal vascular system, resulting in hemangiomatosis. The classic feature of this disease is angioma of the leptomeninges. The other common clinical features are epilepsy (80%), dermal angiomas resulting in portwine stains (76%), abnormal findings in skull radiographs (63%), mental retardation (54%), ocular involvement (37%), and hemiplegia (37%)[2,3].

SWS is classified into three different subtypes by Roach *et al*.[4] In type I (classic) SWS, the individual demonstrates facial angioma, leptomeningeal angioma, and glaucoma; in type II SWS, the patient has facial angioma and glaucoma, with no evidence of intracranial lesions; in type III SWS (rarest variant), the patient presents with only leptomeningeal angioma.[4] The three forms of the syndrome are diagnosed mainly on clinical grounds by the association of the typical cutaneous, central nervous system, and ocular abnormalities.

The incidence of SWS is 3% (three percent) in patients with a facial port-wine hemangioma[5]. The risk of sturge -Weber syndrome is higher when facial haemangiona involves the distribution of the ophthalmic division of the trigeminal nerve. Facial port-wine hemangiomas, whether or not associated with SWS, usually have a sharp midline demarcation, although some extension over the midline has been observed[6].

Glaucoma in this syndrome is almost always unilateral and ipsilateral to the port-wine stain, although contralateral or bilateral glaucoma with unilateral cutaneous lesions have been reported.[7]The occurrence of glaucoma has been noted, especially when the facial skin changes involve the upper and lower eyelids. Numerous mechanisms have been postulated to explain the pathogenesis of glaucoma in SWS. At present, the most accepted explanation for the elevated intraocular pressure is a combination of developmental angle anomalies, which have a dominant role in infantile onset glaucoma and elevated episcleral venous pressure, which is more important in later onset glaucoma[7].

Department of Ophthalmology[1], University of Uyo Teaching Hospital Uyo Nigeria
Department of Ophthalmology[2], Enugu State University Teaching Hospital Enugu Nigeria.

*Corresponding author: favouredchinawa@gmail.com

In the brain, atrophy and calcifications are considered to be an indirect consequence of chronic ischemia of the cortex due to vascular stasis in the area of leptomeningeal angioma[8]. The best modality to demonstrate these vascular malformation, atrophy and calcification of the cortex are Computed Tomography angiography (CTA), Magnetic Resonance Imaging (MRI), and Computed Tomography(CT). Recent advances in neuroimaging have provided important insights into the progression of neurologic injury that occurs as a result of impaired blood flow. Important limitations exist, however, as currently the early diagnosis and exclusion of SWS is impaired by the poor sensitivity of imaging in the newborn period and early infancy[1].

Case Report

We present a 23 year old patient who presented to the clinic with darkening of the right side of the face since childhood, gradual onset of painful blurring of vision in the right eye of 2 years duration and itching of 1 year duration. There was a positive history of redness of the right eye since childhood with subsequent darkening, halos and decreased brightness of light in the right eye associated with the blurring of vision. There was a history of glass cut to his right upper lid about 7 years prior to presentation, with associated redness of the right eye. Symptoms however completely resolved after about 3 weeks of management. There is no family history of similar complaints. There was also no history of seizures, weakness or paralysis of any part of the body. General exam revealed hyperpigmentation of the skin on the right side of the face, over areas supplied by the divisions of the trigeminal nerve (Fig 1).

Ocular exam showed a Snellen visual acuity in the right eye of 6/18 improving to 6/9 with pin hole. He had 6/6 vision in the left eye. Anterior segment findings in the right eye included prominent conjunctival vessels, bluish discoloration of the episclera with underlying tortuous vessels, open anterior chamber angle (Shaffer's grade 3) ,marked angle pigmentation, diffuse and marked pigmentation of the iris, miosed pupil, while posterior segment examination was done with slit lamp biomicroscopy (HergStriet Bern:B90010303 Switzerland) and 78 D. This revealed a pink disc with 0.5 vertical cup disc ratio (VCDR) and increased pigmentation of the retina. Poor dilatation of the pupil in the right eye precluded a fundus photograph. Findings in the left eye were essentially normal, with lesser iris pigmentation, pink disc with 0.3 VCDR and lighter retina. Intraocular pressure (IOP) was assessed with non contact tonometer (NCT tonometer (Topcon) China). IOP in the right eye was 25mmHg while 5mmHg in the left eye. Anisocoria and right iris hyperchromia can be observed in Fig 1. Central visual field of the right eye (Fig 2) showed a seidel's scotoma, in keeping with optic neuropathy and along with raised IOP and cup disc asymmetry, a diagnosis of glaucoma right eye was made.

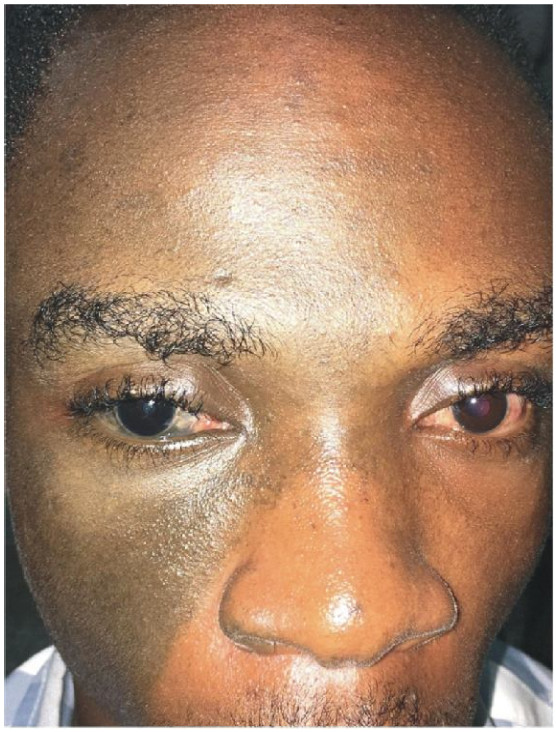
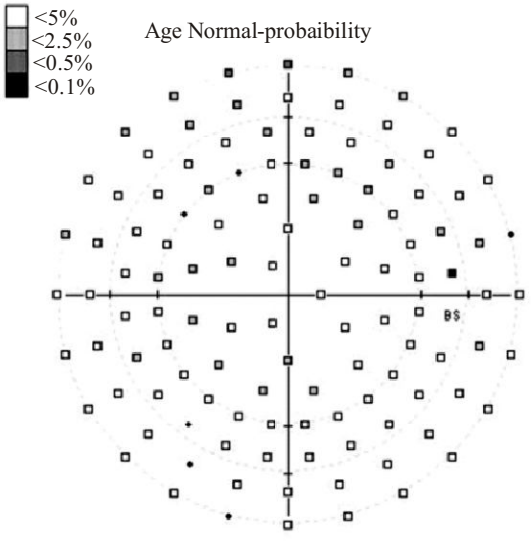


Fig 1: Photo showing altered right-sided naevus flammus, discoloration of right sclera, right iris hyperchromia and anisochoria (worse in dim illumination)



Duration	: 12:08:16 AM	Slope	: 100 dB/10^0*
Lost fixation	: 1/29 (3%) V.good	3 deg. Level	: 21.44 dB
False positives:	1/20 (5%) V.good	Zero level	: 18 dB
False negatives:	0/20 (0%) V.good	Average	: 18.30 dB
Tested	: 591/116	Fluctuation	: -

PD : 1.12 dB	Stimulus : Green III
AD : -8.42dB**	Background : White 3.2 cd/m2
GHT : 0:1; -1; -2; 0 (dB)	
No zone defects	Age norm standard : 2.0
Fovea:-	

Fig 2: CVF plot of the right eye showing a seidel'sscotoma

Combined A-B scan of the right eye (Fig 3) revealed thickening of the choroid, suggesting a choroidalhaemangioma. That for the left eye appeared normal.

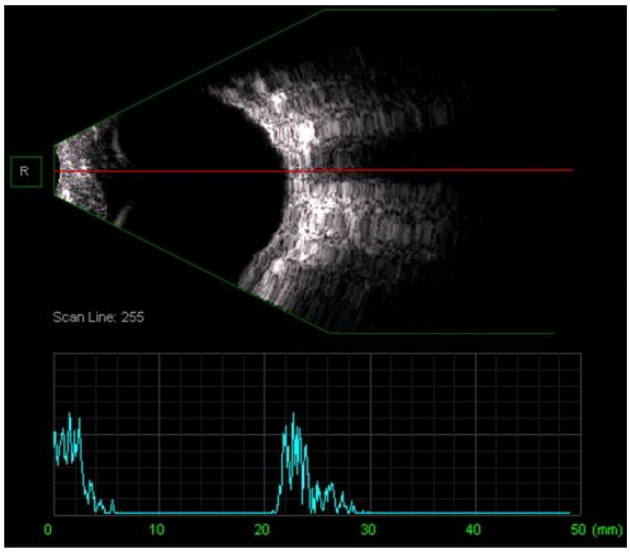


Fig 3: Combined A-B scan if the right eye showing some choroidal thickening

The Patient was refracted and got a best corrected visual acuity of 6/6. He was also started on a combination of a prostaglandin (Latanoprost, 0.005%) and a beta blocker (Timolol, 0.5%) for IOP control in the right eye.

DISCUSSION:

Sturge-Weber syndrome typically presents at birth with facial angiomas. Some authors define SWS as a triad of congenital unilateral port-wine nevus (flat capillary facial angioma) affecting the area innervated by the first sensory branch of the trigeminal nerve, ipsilateral leptomeningeal angiomatosis, calcification in the occipital or frontoparietal region .The third criteria being glaucoma and other vascular eye abnormalities[9]. Our patient presented with Portwine stain, Ipsilateral Glaucoma, iris hyperchromia, episcleral haemangioma and choroidal thickening.

Although Sturge-Weber syndrome appears with a number of manifestations, they are highly variable. In the case presented here, the systemic manifestation was just port wine stains on the face. Vascular lesions in the region innervated by the ophthalmic branch of the

trigeminal nerve is a pointer to central nervous system (CNS) involvement[10]. CNS involvement could not be ascertained in our patient since he was unable to do CT scan or MRI owing to financial constraint. This means he may have had type 1 or 11 SWS.

The facial cutaneous lesion usually is the first component of the syndrome to be observed, since it is visible at birth. Although not medically threatening, it may carry a psychological impact. It may be very pale at first. Although it does not increase in extent, it usually becomes darker with age [11] just as in our patient.

The glaucoma is almost always unilateral and ipsilateral to the port-wine stain, as in our patient. The occurrence of glaucoma has been noted, especially when the facial skin changes involve the upper and lower eyelids which was the case in our patient. Numerous mechanisms have been postulated to explain the pathogenesis of glaucoma in SWS. At present, the most accepted explanation for the elevated intraocular pressure is developmental angle anomalies[7] which may have been demonstrated by marked pigmentation of the angle in our patient. Angle anomaly has a dominant role in infantile onset glaucoma. Another cause of elevated IOP is elevated episcleral venous pressure,[7] which may have been demonstrated by marked tortuosity of episcleral vessel and also darkening of the episclera .

CONCLUSION

Every case of facial angioma in the distribution of trigeminal nerve should have thorough ocular assessment even when there is midline extension of the lesion. This is to rule out Glaucoma which could cause irreversible visual loss.

REFERENCES

1. Comi AM. Update on Sturge-Weber syndrome: diagnosis, treatment, quantitative measures, and controversies. *Lymphat Res Biol* 2007;5:257-64.
2. Gorlin RJ, Pindborg JJ, editors. Syndromes of head and neck. *New York: McGraw-Hill*; 1964;406-9.
3. Caiazzo A, Mehra P, Papagearge MB. The use of preoperative percutaneous transatheter vascular occlusive therapy in the management of sturge weber syndrome-Report of a case. *J Oral Maxillofac Surg*. 1998; 56:775-8.
4. Roach ES. Neurocutaneous syndromes. *PediatrClin North Am*. 1992; 39:591-620
5. Hennedige AA, Quaba AA, Al-Nakib K. Sturge-Weber syndrome and dermatomal facial port-wine stains: incidence, association with glaucoma, and pulsed tunable dye laser treatment effectiveness. *PlastReconstrSurg* 2008; 121:1173-80.
6. Baselga E. Sturge-Weber syndrome. SeminCutan Med Surg 2004; 23:87-98.
7. ArifWahab. Sturge Weber syndrome-a review: *Bombay hospital Journa*l 2008; 1
8. Ling C, Jinjin Wu, Minhui X, Nian Chen and Yadong Y. Sturge-Weber Syndrome.*Ann Dermatol*. 2011; 23:551-3.
9. Pascual-Castroviejo I, Díaz-Gonzalez C, García-Melian RM, Gonzalez-Casado I, Muñoz-Hiraldo E. Sturge-Weber syndrome: Study of 40 patients. *PediatrNeurol*. 1993;9:283-8.
10. Manivannan N, Gokulanathan S, Ahathya RS, Gubernath, Daniel R, Shanmugasundaram. Sturge-Weber syndrome. *Journal of Pharmacy &Bioallied Sciences*. 2012;4(Suppl 2):S349-S352.
11. Mohammed Haneef. Sturge-Weber Syndrome. *Kerala Medical Journal*.2013;6:106-8