

Case Report: A Case of Type II Bilateral Sturge- Weber Syndrome

Dr. K. Sambasivarao ¹, Dr. B. Ushalatha ², Dr. Saradadevi ³.

1. Assistant Professor Radiodiagnosis, Rangaraya Medical College, Kakinada, Andhrapradesh, India

2. Assistant Professor Ophthalmology, Rangaraya Medical College, Kakinada, Andhrapradesh, India

3. Senior Resident, Department of Ophthalmology, Rangaraya Medical College, Kakinada, Andhrapradesh, India

Abstract: Sturge –Weber syndrome (SWS) or encephalotrigeminal angiomas is a sporadic, congenital phakomatosis of unknown etiology. Diagnosed by the triad of facial port wine stain (PWS) in the trigeminal nerve distribution, leptomeningeal venous angiomas, and glaucoma. Bilateral Sturge-Weber Syndrome is a rare entity. We present in this report a rare case of bilateral SWS type II, with PWS involving the Ophthalmic, Maxillary divisions of trigeminal nerves with choroidal hemangiomas and late onset glaucoma but without any neurological affection.

Keywords: Sturge –Weber syndrome (SWS), Facial port wine stain (PWS), Choroidal hemangiomas, Glaucoma.

I. Introduction

Sturge –Weber syndrome (SWS) or encephalotrigeminal angiomas is a non hereditary, non familial, congenital phakomatosis characterised by facial port wine stain vascular nevus flammeus in the trigeminal nerve distribution predominantly involving the Ophthalmic division, leptomeningeal venous angiomas, seizures, dementia, hemiplegia, hemianopia, buphthalmos, and glaucoma. The precise etiology is unknown, but is probably due to faulty development of cortical venous drainage. Bilateral Sturge-Weber Syndrome is a rare entity.

II. Case Report

A 26 year old male attended the outpatient clinic of the Department of Ophthalmology, Government General Hospital, Kakinada, with chief complaints of progressive diminution of vision in both eyes for the past 2 to 3 years. There was history of photophobia, blurring of vision, and occasional pain in both eyes. There was no history of seizures, hemiparesis.

On examination the patient had bilateral capillary hemangioma (PWS) involving the upper one -thirds of face and eyelids on left and entire right half on right (figure.1) described by the patient as a birth mark. The upper lip was protruding and hypertrophied along with gingival hypertrophy. Best corrected visual acuity was 6/60 on right and 6/12 on left. Intraocular pressure measured with applanation tonometer was 36mm Hg in the right eye and 32mmHg in the left eye. Thickening of both upper and lower eyelids with mechanical ptosis in the right upper eyelid.

Anterior segment examination of the both eyes revealed dilated, tortuous conjunctival vessels, clear cornea, with a shallow anterior chamber, papillary reaction was brisk with a clear lens. (figure.2) Fundus examination showed media hazy with coarse vitreous floaters, dilated tortuous vessels with arteriovenous ratio of 2:3, cup-disc ratio of 0.7:1, and Macular edema peripapillary choroidal hemangioma in the right eye (figure.3) and hazy media with coarse vitreous floaters, dilated tortuous vessels with arteriovenous ratio of 2:3, cup-disc ratio of 0.4:1, and Macular edema, and peripapillary choroidal hemangioma in the left eye (figure.4).



Figure.1. PWS involving the right half and upper one-thirds of face and eyelids on left.



Figure.2. Dilated, tortuous conjunctival and episcleral vessels bilaterally.

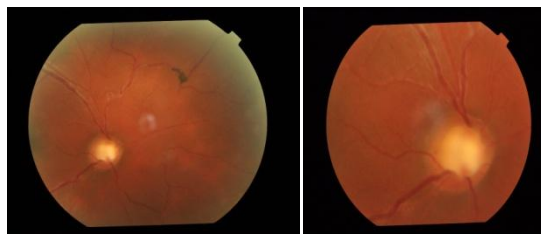


Figure.3. Fundus : Right eye: Choroidal hemangioma with dilated tortuous vessels

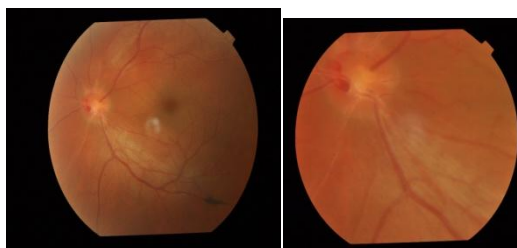


Figure.4. Fundus : Left eye: Choroidal hemangioma with dilated tortuous vessels

Plain CT of the brain revealed normal study with no evidence of any calcifications.

Gadolinium enhanced MRI of the brain(figure.5) and orbits(figure.6) revealed leptomeningeal thickening bilaterally, dilated medullary veins in the right parieto occipital region with enlarged right choroid plexus with thickening and marked enhancement of the posterior wall of the globe bilaterally.

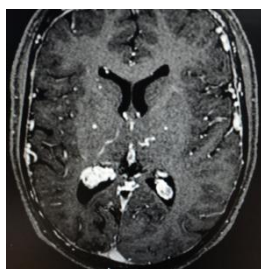


Figure.5. Bilateral leptomeningeal thickening with enlarged right choroid plexus



Figure.6. Thickening and marked enhancement of the posterior wall of the globe bilaterally.

A diagnosis of Type II Sturge-Weber Syndrome was made based on the Cutaneous, Ocular findings and absence of any Neurological signs and symptoms. Topical anti-glaucoma treatment was started to lower the IOP in order to prevent Optic nerve damage.

III. Discussion

Sturge – Weber syndrome (SWS) or encephalotrigeminal angiomatosis is classified by Roach according to the presenting symptoms into three types as Type I with cerebral, cutaneous and ocular manifestations, Type II with only cutaneous and ocular manifestations and Type III with cerebral and cutaneous manifestations 1.

The hallmark of SWS is the presence of facial nevus flammeus or port wine stain in the distribution of Trigeminal nerve predominantly involving the Ophthalmic division which is present in about 98% of the cases. It represents dilated dermal capillaries due to capillary vascular malformation.

The most striking neurological feature of SWS is leptomeningeal angioma ipsilateral to the facial PWS2. It presents as seizures, hemiparesis, developmental delay or mental retardation.

The present case had bilateral nevus flammeus involving the first division of trigeminal nerve on left and all branches of trigeminal nerve on right . The upper lip was protruding and hypertrophied along with gingival hypertrophy but without any neurological impairment.

Gadolinium enhanced MRI Brain is the Gold standard for diagnosing SWS3. In our case MRI brain with orbits revealed leptomeningeal thickening, dilated medullary veins in the right parieto occipital region with enlarged ipsilateral choroid plexus with thickening and marked enhancement of the posterior wall of the globe bilaterally.

Ocular abnormality is more commonly seen in patients with PWS in the first and second divisions of trigeminal nerve4. Glaucoma is a prominent feature of SWS. Bilateral glaucoma is seen in about 45% of patients of bilateral SWS. Choroidal and episcleral hemangiomas are other ocular findings commonly found in patients of SWS. The episcleral vessel tortuosity, probably resulting from arteriovenous shunts within the episcleral hemangiomas, represents raised episcleral venous pressure that is implicated in causation of late onset glaucoma. Facial nevus flammeus involving the palpebral area is a strong indicator of choroidal hemangioma5. Dilated and tortuous retinal vessels with peripheral retinal arteriovenous communications may also be visible on fundus examination.

The episcleral telangiectasia was seen in both the eyes with bilateral open-angle glaucoma in this case, who presented quite late to us. Choroidal hemangiomas were noted in both the eyes on fundus examination which was confirmed on Gadolinium enhanced MRI of orbit. There were dilated tortuous retinal vessels in both eyes with arteriovenous communication in periphery of retina.

Gingival involvement is reported in 40% cases of SWS. The oral lesions include gingival hemangioma, gum hypertrophy, and asymmetric jaw growth6.

To summarize, the current case of type II SWS presented with bilateral nevus flammeus involving the first and second divisions of trigeminal nerve and third division on right with gingival hypertrophy but without any neurological impairment. Ocular findings include bilateral episcleral telangiectasia, open angle glaucoma, Choroidal hemangioma, retinal arteriovenous malformations. Although there was no clinical evidence of neurological deficit, Gadolinium-enhanced MRI of brain showed leptomeningeal thickening, dilated medullary veins in the right parieto occipital region with enlarged ipsilateral choroid plexus.

In conclusion, the absence of classical triad of central nervous system, ocular, and cutaneous involvement does not preclude the diagnosis of SWS. Furthermore, SWS may present with incomplete and atypical presentation as highlighted in the reported case8.we conclude ocular examination should be a mandatory for all cases with facial nevus flammeus as it would predict the future ophthalmic complications at an earlier stage.

References

- [1]. Roach ES. Neurocutaneous syndromes. *Pediatr Clin North Am* 1992;39:591-620.
- [2]. Zhou J, Li NY, Zhou XJ, Wang JD, Ma HH, Zhang RS. Sturge-Weber syndrome: A case report and review of literatures. *Chin Med J* 2010;123:117-21.
- [3]. Pascual-Castroviejo I, Pascual-Pascual SI, Velazquez-Fragua R, Viano J. Sturge-Weber syndrome: Study of 55 patients. *Can J Neurol Sci* 2008;35:301-7.
- [4]. Govori V, Gjikolli B, Ajvazi H, Morina N. Management of patient with Sturge-Weber syndrome: A case report. *Cases J* 2009;2:9394
- [5]. Del Monte MA. Sturge-Weber Syndrome. [Electronic version]. Available from: <http://emedicine.medscape.com/article/1219317-overview>.
- [6]. Thomas-Sohl KA, Vaslow DF, Maria BL. Sturge-Weber syndrome: A review. *Pediatr Neurol* 2004;30:303-10.
- [7]. Mukhopadhyay S. Sturge-Weber syndrome -A case report. *J Indian Soc PedodPrev Dent* 2008;26:29-31
- [8]. Nitin N, Jagriti J. A rare presentation of bilateral sturge weber syndrome, *Oman j of Ophthal* 2014;7: 46-48
- [9]. Tzoukeva Al., N. Deleva, localized choroidal haemangioma associated with sturge – weber syndrome: a case report. *Journal of IMAB* 2008, book 1 95-97.