Case Report

Weber syndrome with secondary glaucoma: A case report

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Abstract

Sturge-Weber syndrome (SWS) is a non-hereditary congenital disorder due to somatic mosaic mutations in the GNAQ gene. The classical presentation relates to the brain lesion (cerebral angiomatous lesion of leptomeninges, which is responsible for epileptic seizures, hemiparesis and mental retardation), skin lesion (unilateral facial nevus), ocular and oral involvement. Patients of SWS are at risk of developing secondary glaucoma. We present here a 9-year-old boy who has port wine stain with secondary glaucoma.

Key Words: Sturge-Weber syndrome; Secondary Glaucoma; Port wine stain

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INTRODUCTION

Sturge-Weber Syndrome (SWS) belongs to a group of disorders known as phacomatosis. Unlike other phacomatosis, SWS has no hereditary pattern and is caused by somatic mosaic mutations in the GNAQ gene.[1] This neurocutaneous disorder characterized by angiomas that involve the leptomeninges (leptomeningeal angiomas) and the skin of the face, typically in the ophthalmic (V1) and maxillary (V2) distributions of the trigeminal nerve. The hallmark of SWS is a facial cutaneous venous dilation, also referred to as a nevus flammeus or port-wine stain (PWS)[2,3]. Ocular manifestations are due to vascular abnormalities of the eyelid, orbit, conjunctiva, episclera, ciliary body, choroid and retina [4]. Patients of SWS are at risk of developing secondary glaucoma[5].

CASE REPORT

A 9-year-old boy presented with left-sided facial nevus since birth. He was born at term by spontaneous vaginal delivery without postnatal complications. He started to walk with limping by the age of 2 years. His cognition was normal and he had good school performance. Physical examination showed a port wine stain involving the left upper eyelid and left side of the face. The rest of systemic examination was unremarkable. In ocular examination, there was increased vascularity of conjunctiva and episclera with crock-screw vessels in left eye, suggesting underlying episcleral hemangiomas. Fundus examination revealed cup disc ratio in R/E- 0.2 and L/E -0.4. The intraocular pressure (IOP) was high (> 30 mm of Hg) in left eye recorded on three occasions. On gonioscopy, the angle structures appear distinct with iris processes in 360 degree. Visual fields and RNFL thickness on OCT were within normal limits. Dental assessment revealed bilateral gingival overgrowth, poor oral hygiene with multiple dental caries. Patient was started on anti glaucoma medication initially on eye drop Timolol 0.25% one drop twice a day in L/E with punctal occlusion and kept under follow up. His IOP was still on higher side (25mm of Hg), so newer drug (eye drop Dorzolamide 2% one drop twice a day) was added to achieve target IOP.

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Figure 1: Facial port wine stain and ocular involvement in 9-year- old patient with Sturge-Weber syndrome.

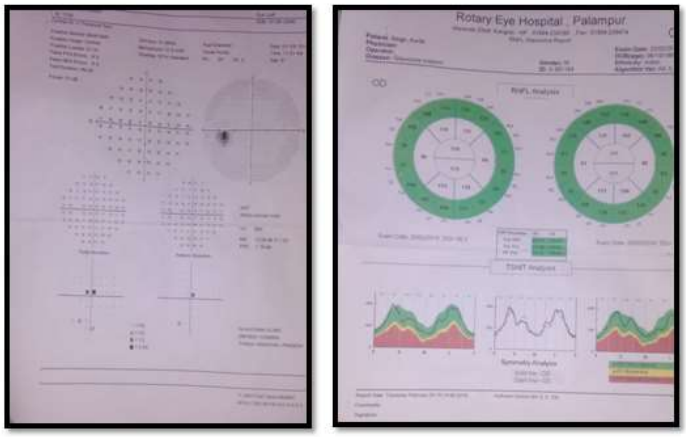

Figure 2: (A) Visual Fields. (B) OCT –RNFL thickness

DISCUSSION

Sturge-Weber syndrome is a congenitally occurring phakomatosis with the pathognomonic finding of unilateral, diffuse, dermal angioma[6]. Other frequent findings include meningeal angioma with tram-track calcifications of the brain[7], choroidal hemangioma, and a unilateral glaucoma on the affected side that more often occurs when the angioma involves the upper lid. Port wine stain is the most common clinical manifestation of SWS, usually presenting unilaterally, typically on the forehead and upper eyelid, and it may be extended to the neck and other parts of the body[8]. This is similar to findings in the present case with port wine stain involving the left upper eyelid and left side of face. The most common ocular presentation in SWS is glaucoma (increased intraocular pressure), which occurs in 30-70% of affected patients[9.10]. When present, elevated IOP develops in infancy or the late teens and can be severe. In newborns, the glaucoma mechanism is more likely to be caused by abnormal trabecular microanatomy similar to that seen in congenital glaucoma, whereas the later-onset form can result from elevated episcleral venous pressure (EVP)[11]. Phelps reported an elevation in EVP related to the extent of the episcleral angioma[12]. Sometimes, this episcleral angioma is not obvious until the conjunctiva and thick Tenon capsule are reflected during surgery. Topical pharmacotherapy (beta-blockers and carbonic anhydrase inhibitors) may normalize the intraocular pressure, yet the majority of patients require surgical management[5]. But standard filtration methods carry a high risk of expulsive choroidal haemorrhage, which may be even more likely to occur in those cases of SWS that have a choroidal hemangioma[13]. The primary cerebral lesion in SWS is the leptomeningeal capillary-venous malformation (leptomeningeal angioma), which is usually located unilaterally over the posterior temporo-parietal and occipital areas, but can occur without an associated facial nevus [14]. Ischemic changes in the tissue surrounding the lesion with unilateral parietal and occipital lobe gyriform calcification lead to convulsions, hemiparesis and cognitive dysfunction[5].

CONCLUSION

SWS is rare, sporadic disorder involving vasculature in the facial skin, CNS, and eye. The chief ocular manifestation is glaucoma in which underlying mechanisms are development anomaly of the anterior chamber angle and elevated episcleral pressure. Such patients require continued vigilance and long term follow up that includes routine workup, ophthalmology assessment, dental assessment if clinically suggested, treatment of seizure and glaucoma. Early diagnosis and intervention may help in preventing, or early treating, further neurological and ocular complications.

REFERENCES

1. Shirley MD, Tang H, Gallione CJ, Baugher JD, Frelin LP, Cohen B, *et al*. Sturge-Weber syndrome and port-wine stains caused by somatic mutation in GNAQ. N Engl J Med 2013; 368:1971-1979.
2. Comi AM. Presentation, diagnosis, pathophysiology, and treatment of the neurological features of Sturge- Weber syndrome. Neurologist 2011; 17:179-184.
3. Sujansky E., Conradi S. Sturge-Weber syndrome: age of onset on seizures and glaucoma and the prognosis for affected children. J Child Neurol 1995;10:49–58.
4. Abdolrahimzadeh S, Scavella V, Felli L, Cruciani F, Contestabile MT, Recupero SM. Ophthalmic Alterations in the Sturge-Weber Syndrome, Klippel-Trenaunay Syndrome, and the Phakomatosis Pigmentovascularis: An Independent Group of Conditions? BioMed Research International. 2015;2015:786519.
5. Budenz DL *et al*. Ophrhalmology. 2000; 107(11):2105-10.
6. Hassan S, Babiker A, Bashiri FA, Hassan H H, Elhusseini M, Salih MA. Sturge-Weber syndrome: Continued vigilance is needed. Sudan J Paediatr 2015; 15(2): 63 - 70.
7. Pascual-Castroviejo I, Díaz-Gonzalez C, García-Melian RM, Gonzalez-Casado I, Muñoz-Hiraldo E. Sturge-Weber syndrome: study of 40 patients. Pediatr Neurol 1993; 9:283-288.
8. Tallman B, Tan OT, Trainor S, Morelli JG, Weston WL, Piepenbrink J, Stafford TJ. Location of port-wine stains and the likelihood of ophthalmic and/or central nervous system complications. Pediatrics 1991; 87:323-327.
9. Sullivan TJ, Clarke MP, Morin JD. The ocular manifestations of the Sturge-Weber syndrome. J Pediatr Ophthalmol Strabismus 1992; 29:349-356.
10. Baselga E. Sturge-Weber syndrome. Semin Cutan Med Surg 2004; 23:87.
11. Weiss DI. Dual origin of glaucoma in encephalotrigeminal haemangiomatosis. Trans Ophthalmol Soc UK. 1973;93(0):477-493.
12. Phelps CD. The pathogenesis of glaucoma in Sturge-Weber syndrome. Ophthalmology. 1978;85(3):276-286.
13. Bellows AR, Chylack LT Jr, Epstein DL, Hutchinson BT. Choroidal effusion during glaucoma surgery in patients with prominent episcleral vessels. Arch Ophthalmol. 1979;97(3):493-497.
14. Aydin A, Cakmakçi H, Kovanlikaya A, Dirik E. Sturge-Weber syndrome without facial nevus. Pediatr Neurol 2000; 22:400-402.

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