Case Report

Sturge-Weber Syndrome: a case report

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ABSTRACT

Sturge-Weber syndrome is a rare congenital, non-familial sporadic condition also known as encephalotrigeminal angiomatosis, is a phakomatoses consisting of neurological, skin and ocular manifestations. Symptoms reported varies in severity and age of onset but it is commonly detected in infancy. The Ophthalmic feature of this disease consists of glaucoma and is also associated with vascular manifestation of the conjunctiva, epi-sclera, choroid and retina. In the present case, a 19-year-old male presented with a port wine stain on the right side of the face, leptomeningeal angioma, ocular hypertension, temporal hemianopia of the left eye and choroidal hemangioma with macular scar of the right eye, seizure and weakness of the left side of the body. The purpose of presenting the case is to illustrate both the characteristic presentation and to underline the importance of its diagnosis in the clinical ophthalmological practice.

Key words: *Sturge-Weber syndrome, glaucoma, port-wine stain, nevus flammus.*

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INTRODUCTION

Sturge-Weber Syndrome (SWS) is a congenital, non-hereditary illness that impacts the brain, eyes and skin. It is also known as encephalotrigeminal angiomatosis. SWS is believed to be caused by the persistence of vascular plexus around the cephalic portion of the neural tube. This plexus develops during the sixth week of intrauterine development but normally undergoes regression during the ninth week.[1] Schirmer reported the first case, although he failed to identify the neurological condition. William Allen Sturge provided a more detailed description of the condition in 1879 and linked the disease's ocular and dermatological alterations to neurological symptoms. In 1929, Frederick Parkes Weber clarified the radiologic characteristics observed in SWS patients.[2]

The most common characteristic that is usually present is the presence of port wine stain (reddish-purple or dark red color) in the ophthalmic and maxillary division of the trigeminal nerve.[3] Additionally, ipsilateral gyri form calcification and venous angiomas in the leptomeninges over the cerebral cortex may exist. These conditions might result in epilepsy and hemiparesis on the opposite side.[4] The most common ocular manifestation is glaucoma and vascular anomalies involving conjunctiva, epi-sclera, choroid and retina. This paper describes a patient who had SWS and displayed some of its characteristic clinical traits. The significance of a precise diagnosis and treatment plan in clinical ophthalmology practice is underlined.

CASE REPORT

A 19-year-old male presented with the complaints of dimness of vision in his right eye and headache since childhood. Physical examination revealed reddish discoloration in right half of face (Figure 1). Visual acuity in the right eye was hand movement and the left eye 6/6. Anterior chamber was normal in depth in both eyes. Conjunctival and episcleral hemangioma were present on the right eye (Figure 2). Intraocular pressure (IOP) on Right eye: 26 mmHg & Left eye 16 mmHg on

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Goldmann Applanation Tonometry(GAT). Gonioscopy revealed grade -3 open angle on both eyes. Fundus examination revealed C:D ratio: 0.3:1 with healthy neuro-retinal rim on both eyes and choroidal hemangioma in superior temporal arcade with macular scar on the right eye and left eye was normal (Figure 3). *Humphrey visual field* analysis (HVFA) (24-2) on left eye revealed temporal hemianopia (Figure 4). Magnetic Resonance Imaging (MRI) of the brain with contrast showed right sided hemi atrophy with leptomeningeal contrast enhancement of right cerebral hemisphere, abnormally large vein in sub ependymal, periventricular region (Figure 5).

On query, patient gave history of convulsion, left sided weakness and port wine stain on the right half of the face since birth. Patient was on regular anticonvulsant drug. There was no history of mental retardation. The patient was also referred to the neurology department for further neurological evaluation.

Figure 1. Port wine stain

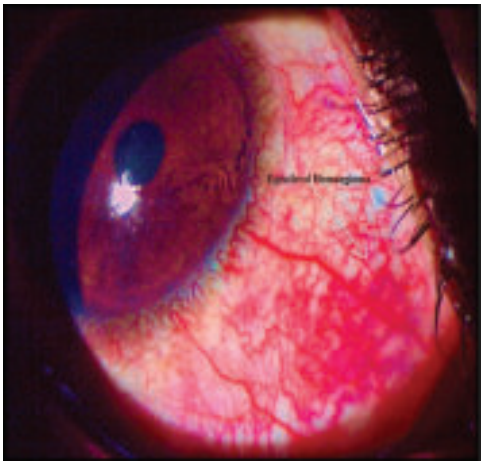

Figure 2. Episcleral hemangioma

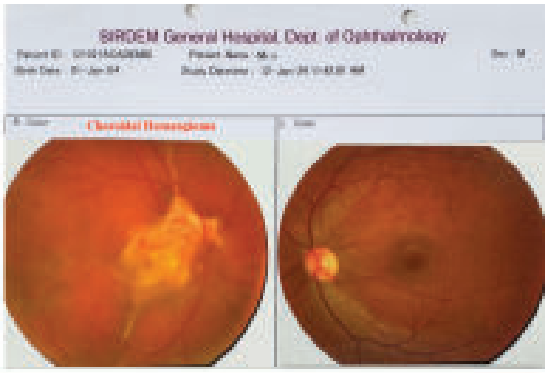

Figure 3. CFP showing choroidal hemangioma RE with normal LE

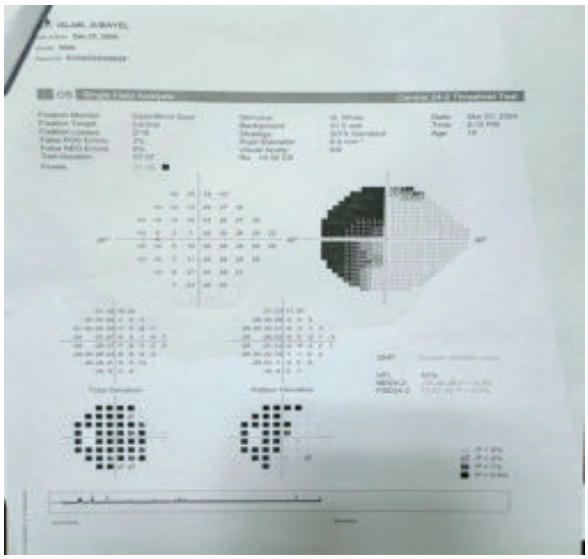

Figure 4. HVFA- LE (Temporal Hemianopia)

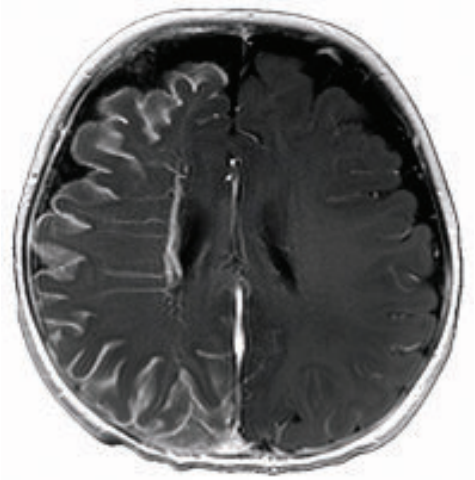

Figure 5. MRI showing Leptomeningeal contrast enhancement

DISCUSSION

SWS has extremely varied clinical features and it can be characterised by Port Wine stain in the face, leptomeningeal angiomas, ipsilateral gyri form calcifications, convulsive crisis, varied forms of ocular involvement & hemi paresis. SWS is referred to as complete when both CNS and facial angiomas are present and incomplete when only one area is affected without the other. The Roach Scale is used for classification, as follows[5]:

Type I - Both facial and leptomeningeal angiomas; may have glaucoma

Type II - Facial angioma alone (no CNS involvement); may have glaucoma

Type III - Isolated leptomeningeal angioma; usually no glaucoma.

According to the above criteria, our case is a complete Type I SWS case.

It has been postulated that the occurrence of leptomeningeal and facial angiomata suggests the persistence of undeveloped sinusoidal vascular channels.[3] The relative deficiency of superficial cortical veins causes shunting (steal) of blood to the deep venous system by the enlarged medullary veins which eventually results in stasis and ischemia. The leptomeningeal angioma, mainly unilateral and located in the occipital or posterior parietal lobe, may initially be unnoticed. However, during the first years of life, impaired venous outflow may lead to progressive ischaemic cerebral damage, atrophy and calcification in the brain region below the angioma.[6] These cerebral manifestations, which can be confirmed by neuroimaging (CT or MRI), regularly cause neurological problems such as seizures (72–97%), motor deficits, cognitive decline and visual field (VF) defects.[7] In our case, MRI of brain with contrast showed right sided hemi atrophy with leptomeningeal contrast enhancement of right cerebral hemisphere, abnormally large vein in sub ependymal, periventricular region.

There is a report of development of raised IOP and glaucoma in 30–70%of SWS cases. Glaucoma shows a bimodal peak of age development. One is early-onset (congenital) form affecting ~60% of patients and another peak is a later-onset form affecting children and adolescence (40% of cases).[8] In our case, ocular hypertension developed and it is unilateral. The posterior segment of the eye is also involved with hemangioma of the choroid (20%–70% of cases).[9] In our case choroidal hemangioma with macular scar was seen on the fundus of the right eye while left eye was normal.

There are many new hypotheses and factors on the pathogenic mechanisms that can lead to increase Intra-ocular pressure (IOP) and the development of glaucoma. The formulated theories are-

A rise in episcleral venous pressure (EVP) because of arteriovenous shunts mechanism into the episcleral hemangioma. This theory is based on the observation of a normal angle structure, blood within Schlemm's canal, and more severe glaucoma and it is mainly responsible for the adult onset glaucoma.[10]

Hypersecretion of fluid either by the ciliary body or the choroidal hemangioma. A variation from normal hemodynamics of the anterior chamber angle and episclera because of premature aging of the trabecular meshwork–Schlemm's canal complex in later-onset glaucoma.[8]

In our case, it can be attributed that the cause of elevated IOP is because of elevated episcleral venous pressure as postulated by Weiss as the main cause of late onset glaucoma.[10]

Tannous et al in their study reported that in late-onset glaucoma, the increase in episcleral venous pressure might be due to progressive hypertrophy and dilatation of the episcleral veins. The use of antiglaucoma drugs like beta-blockers and carbonic anhydrase inhibitors is found to be effective in some SWS patients with absence of buphthalmos in a few case reports.[11] In our case, a combination of beta blocker and carbonic anhydrase inhibitors was used to lower the IOP.

Facial port wine stains can be successfully treated by laser. Laser should be done superficially to avoid potential complications like decrease in brain venous outflow through PWS vessels, leading to deterioration of cerebral veins, dilatation of choroidal vessels, exudative retinal detachment, and rise in IOP. Therefore, deep photocoagulation and debulking surgery should be avoided in the treatment of PWS.[11]

Conclusion

SWS is a rare congenital disease that has classical symptomatology and neurological findings. A

multidisciplinary strategy is employed to achieve the symptom-driven therapy goal. The most frequent eye consequence is glaucoma. Though in our case, poor vision in right eye was due to cerebral atrophy of same side of the brain & choroidal hemangioma with macular scar, we also started topical anti-glaucoma medication to prevent further visual loss due to ocular hypertension. In fact, glaucoma in SWS which is early onset, is poor response to conventional medical therapy, surgery is frequently necessary to achieve long-term control of the condition.

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Consent: Informed written consent was obtained from the patient and his attendant for the publication of case report and its image.

Conflicts of interest: Nothing to declare.

REFERENCES

1. Bachur CD, Comi AM. Sturge-weber syndrome. Curr Treat Options Neurol 2013;15(5):607-17.
2. Neville BW, Damm DD, Alen CM, Boquot JE. Oral and Maxillofacial Pathology. 2nd ed. Philadelphia: Elsevier; 2002. p. 471-3.
3. Waelchli R, Aylett SE, Robinson K, Chong WK, Martinez AE, Kinsler VA. New vascular classification of port-wine stains: Improving prediction of Sturge–Weber risk. Br J Dermatol 2014;171(4):861-7.
4. Khambete N, Risbud M, Kshar A. Sturge-Weber syndrome: a case report. Int J Dent Clin 2011;3(1):79.
5. Royle HE, Lapp R, Ferrara ED. The Sturge-Weber syndrome. Oral Surgery, Oral Medicine, Oral Pathology 1966; 22(4):490-7.
6. Jeong JW, Tiwari VN, Shin J, Chugani HT, Juhász C. Assessment of brain damage and plasticity in the visual system due to early occipital lesion: comparison of FDG-PET with diffusion MRI tractography. J Magn Reson Imaging 2015 Feb;41(2):431-8.
7. Zabel TA, Reesman J, Wodka EL, Gray R, Suskauer SJ, Turin E, et al. Neuropsychological features and risk factors in children with Sturge-Weber syndrome: four case reports. Clin Neuropsychol 2010;24(5):841-59.
8. Sharan S, Swamy B, Taranath DA, Jamieson R, Yu T, Wargon O, et al. Port-wine vascular malformations and glaucoma risk in Sturge-Weber syndrome. J Am Assoc Pediatr Ophthalmol Strabismus 2009;13(4):374-8.
9. Tannous Z, Rubeiz N, Kibbi AG. Vascular anomalies: portwine stains and hemangiomas. J Cutaneous Pathol 2010; 37:88-95.
10. Basler L, Sowka J. Sturge-Weber syndrome and glaucoma. Optometry 2011;82(5):306-9.
11. Parsa CF. Focal venous hypertension as a pathophysiologic mechanism for tissue hypertrophy, port-wine stains, the Sturge-Weber syndrome, and related disorders: proof of concept with novel hypothesis for underlying etiological cause (an American Ophthalmological Society thesis). Transactions of the American Ophthalmological Society 2013;111:180.