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VISION UNDER SIEGE: A CASE OF STURGE-WEBER SYNDROME UNVEILING GLAUCOMA

Ophthalomology

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ABSTRACT

Sturge-Weber syndrome (SWS) is a congenital disorder belonging to a group called phakomatoses involving hamartomas that affect the skin, eyes, and brain, leading to symptoms like facial port-wine stain, glaucoma, choroidal vascular malformations, and cognitive issues. Patients with SWS frequently experience glaucoma. This report focuses on a 15-year-old male with SWS who had a port-wine stain on the right side of his face and glaucoma on the same side. The case illustrates the need for early and ongoing monitoring of eye and brain symptoms in individuals with SWS. Timely intervention can help prevent complications and improve patient outcomes, demonstrating the importance of a multidisciplinary treatment approach for managing SWS.

KEYWORDS

Sturge Weber syndrome, glaucoma, neurocutaneous disorder, seizures

INTRODUCTION:

Sturge Weber Syndrome (SWS) is a neurocutaneous condition that features capillary venous malformations in the skin, eye, and brain. [1] It ranks third in frequency among neurocutaneous syndromes.

Key clinical features include a unilateral facial port-wine stain (PWS) in areas supplied by the ophthalmic and maxillary branches of the trigeminal nerve and abnormal capillary development in the choroid and brain, leading to varied neurological and ocular manifestations.

Roach et al. classify SWS into four subtypes. In type I SWS, individuals exhibit facial angioma and leptomeningeal angioma, with or without glaucoma. Type II SWS involves facial angioma, with or without glaucoma, but lacks intracranial lesions. Type III SWS presents only with leptomeningeal angioma; Type IV presents with systemic manifestations along with type I features. [2]

Ocular findings are observed in about half of SWS patients. Glaucoma develops on the same side as the PWS in 30%–70% of SWS cases. [3] The development of glaucoma is intricate, involving congenital abnormalities in the anterior chamber angle that hinder the outflow of aqueous humor. This condition can be exacerbated by elevated episcleral venous pressure, increased production of aqueous humor by the ciliary body or choroidal hemangioma, and unusual hemodynamics in the episcleral and anterior chamber angles due to early aging of the trabecular meshwork and Schlemm's canal complex. [4]

This article documents the case of a 15-year-old male with SWS who presented with glaucoma and episcleral congestion.

Case report:

A 15-year-old male from a lower socioeconomic background presented with a gradual, progressive, and painless decrease in vision in his right eye over the past five years. He also reported redness in his right eye for five years and had been experiencing seizures since the age of four. The patient's attendant also gave a history of pigmentation on his right side of the face since birth. [Figure 1] Patient seizures were controlled on antiepileptic medications. There was no history of ocular trauma, surgery, systemic illness, medication use, or significant family history.

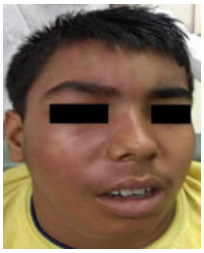

Figure 1: Port wine stain on right side of face

Upon general examination, a port-wine stain was noted on the right side of his face, including the forehead, nose, cheeks, and upper lip up to the angle of the mouth. Anterior segment ocular examination of both eyes was within normal limits except for episcleral vessel congestion, which was observed in the right eye. [Figure 2]

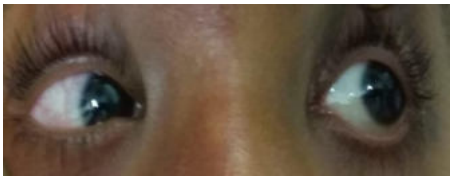

Figure 2: Episcleral vessel congestion in the right eye

The best-corrected visual acuity was 6/18 in the right eye and 6/6 in the left eye. Intraocular pressure (IOP) measured 38 mmHg in the right eye and 18 mmHg in the left eye. Fundus examination showed glaucomatous changes with CD ratio 0.9:1 in the right eye, while the vessels and foveal reflex appeared normal. The left-eye fundus was normal. [Figure 3] Gonioscopy of both eyes showed a wide-open angle.

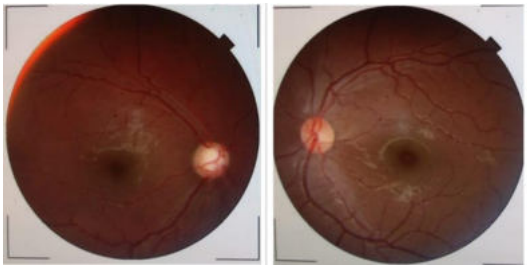

Figure 3: Fundus photograph of both eyes showing CD Ratio of 0.9: 1 in the right eye

Neurological examination revealed left hemiparesis, and the results of the neuropsychological evaluation indicated borderline to mildly impaired cognitive performance.

A non-contrast CT (NCCT) scan of the head revealed extensive curvilinear calcifications in the right temporoparietal occipital region, with no involvement of white matter, along with atrophic changes in the right brain parenchyma, indicative of Sturge-Weber syndrome. [Figure 4] The patient was started on antiglaucoma medications (Bimatoprost 0.01%, dorzolamide 2%, and timolol 0.5%) and was advised to follow up regularly.

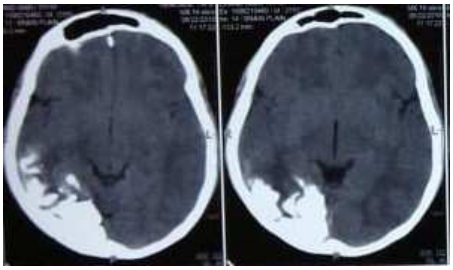

Figure 4: Non-contrast CT scan of the head revealed extensive curvilinear calcifications in the right temporoparietal occipital region

DISCUSSION:

Sturge-Weber syndrome (SWS) is an uncommon sporadic disorder that impacts multiple organs. Although capillary malformation (CM) is believed to contribute to the syndrome's manifestations, the precise pathogenesis remains unknown. Recent studies have found the guanine nucleotide-binding protein G(q) subunit alpha (GNAQ) R183Q mutation in 90% of SWS cases [5], indicating that this mutation may cause CM by stimulating endothelial cell proliferation. [6] The somatic GNAQ mutation is predominantly located in the endothelial cells of capillary malformations in the skin, brain, and choroid. The precise impact of this mutation on these cells is still not fully understood. Other theories suggest the continued presence of primordial sinusoidal vascular channels and alterations in perivascular vessel innervation due to neural crest cell abnormalities.[7]

Epilepsy, hemiparesis, intellectual disabilities, and ocular issues are among the most frequent and severe symptoms in patients with SWS. These patients often face challenges in integration due to limited cognitive and social skills, poor aesthetic appearance, and epilepsy. The neurological symptoms of SWS are thought to result from vascular stasis and poor perfusion in the cortex beneath the leptomeningeal capillary malformation. Additionally, disruption of the hypothalamic-pituitary axis by SWS can lead to endocrine issues, such as growth hormone deficiency and hypothyroidism. [8]

Glaucoma is a common ocular manifestation in SWS, affecting 30–70% of patients. It typically presents as open-angle glaucoma, often accompanied by other findings like buphthalmos, and has two peaks of onset: early (congenital) and later in childhood or adolescence. Choroidal hemangiomas, which occur in 20–70% of SWS patients, can increase the risk of glaucoma. Although often asymptomatic, severe choroidal hemangiomas can lead to visual impairment through exudative retinal detachment and macular edema. [9]

In this case, the patient exhibited ocular abnormalities and cutaneous nevus flammeus, leading to a diagnosis of SWS. His history of progressive, painless vision loss in the right eye, unilateral redness since childhood, and seizures beginning at age five is consistent with typical SWS manifestations.

This report emphasizes the critical need for early detection and management of SWS-related glaucoma to prevent permanent vision loss. The patient was promptly started on antiglaucoma medications and advised to have regular follow-ups, underscoring the necessity for continuous monitoring and intervention to effectively manage this complex condition. Given the risk of severe outcomes, including frequent strokes, tonic or myoclonic seizures, and progressive visual impairment, a multidisciplinary approach involving neurology, ophthalmology, and dermatology is crucial for comprehensive care for SWS patients.

CONCLUSION:

This case highlights the critical importance of early detection and ongoing management of glaucoma and other ocular and neurological manifestations in patients with Sturge-Weber Syndrome (SWS). The complex nature of SWS, involving multiple organ systems, underscores the necessity for a multidisciplinary approach, integrating neurology, ophthalmology, and dermatology, to provide comprehensive care and improve patient outcomes. This case emphasizes that vigilant monitoring and a proactive treatment strategy can significantly enhance the quality of life for individuals with SWS.

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