ISSN: 2581 – 3633
PubMed - National Library of Medicine - ID: 101745081

International Journal of Medical Science and Advanced Clinical Research (IJMACR)

Available Online at:www.ijmacr.com

Volume – 8, Issue – 5, October - 2025, Page No.: 16 – 22

Sturge–Weber Syndrome: A Case Series of Five Patients Emphasizing Ocular and Psychiatric Manifestations

[1]Dr Vipin Kumar, Associate Professor, Department of Psychiatry, SHKM, GMC, NUH

[2]Dr Neha Adlakha, Associate Professor, Department of Ophthalmology, SHKM, GMC, NUH

[3]Dr (Lt Col) Abhishek Bharti, Professor Psychiatry, MMIMSR, Mullana, Ambala, Haryana

Corresponding Author: Dr Neha Adlakha, Associate Professor, Department of Ophthalmology, SHKM, GMC, NUH.

How to citation this article: Dr Vipin Kumar, Dr Neha Adlakha, Dr (Lt Col) Abhishek Bharti, "Sturge–Weber Syndrome: A Case Series of Five Patients Emphasizing Ocular and Psychiatric Manifestations", IJMACR- October - 2025, Volume – 8, Issue - 5, P. No. 16 – 22.

Type of Publication: Case Series

Conflicts of Interest: Nil

Abstract

Background: Sturge–Weber syndrome (SWS) is a rare neurocutaneous disorder characterized by facial capillary malformation (port-wine stain), leptomeningeal angioma, and ocular vascular anomalies. Though neurological features like seizures are well described, the ocular and psychiatric manifestations are less often systematically reported.

Methods: We present a case series of five patients with SWS, each with detailed ophthalmologic evaluation and psychiatric assessment. We review ocular findings (glaucoma, choroidal hemangioma, retinal changes) and psychiatric comorbidities (mood disorders, psychosis, cognitive impairment) and discuss their management.

Results: All five had characteristic facial port-wine stains and neuroimaging evidence of leptomeningeal involvement. Ocular involvement ranged from congenital glaucoma, adult-onset glaucoma, diffuse choroidal hemangioma, and secondary retinal changes. Psychiatric manifestations included mood disorders, psychosis, anxiety, and intellectual disability. Outcomes varied depending on severity and timing of intervention.

Conclusions: Multidisciplinary surveillance including ophthalmology and psychiatry is important in SWS. Recognition of psychiatric burden is essential for comprehensive care.

Keywords: Glaucoma, Psychosis, Neurologically Sturge–Weber Syndrome

Introduction

Sturge–Weber syndrome (SWS), also known as encephalotrigeminal angiomatosis, is a sporadic neurocutaneous disorder caused by postzygotic somatic mutations in the GNAQ[1] gene. This leads to abnormal development of vascular structures in the brain, skin, and eyes. SWS is characterized by a triad of facial port-wine stain, leptomeningeal angiomas, and ocular vascular anomalies[2]. Historically, the condition was classified among phakomatoses but is now recognized as a somatic

mosaic disorder, explaining its segmental distribution and sporadic occurrence.

The syndrome manifests with a wide spectrum of neurological, ocular, dermatologic, and psychiatric features. Neurologically, SWS often presents with seizures, hemiparesis, developmental delays, and stroke-like episodes. Early-onset seizures are associated with more severe cognitive impairment and can be refractory to standard antiepileptic therapies[3]. Neuroimaging typically reveals cortical atrophy, gyriform calcifications, and leptomeningeal angiomas, most commonly in occipital and posterior parietal regions. Ocular manifestations are frequent and diverse. Glaucoma is the most common complication, occurring either congenitally or later in life due to trabeculodysgenesis or elevated episcleral venous pressure. Diffuse choroidal hemangiomas may result in visual loss from serous retinal detachment[4]. Other ocular findings include optic nerve atrophy, retinal vascular tortuosity, and refractive errors. Early diagnosis and intervention are critical to preserve vision. Psychiatric and cognitive complications, though less studied, significantly impact quality of life. Patients may exhibit intellectual disability, attention deficits, behavioral disorders such as ADHD, mood disorders including depression and anxiety, and rarely, psychotic features. The interplay of chronic epilepsy, cortical vascular anomalies, and psychosocial stress contributes to these neuropsychiatric outcomes[5].

Classification of SWS, according to Roach et al., includes Type I (facial and leptomeningeal angiomas, may include glaucoma), Type II (facial angioma alone, with or without glaucoma), and Type III (isolated leptomeningeal angioma without facial lesions). Type I is most commonly associated with ocular and psychiatric manifestations.

Understanding the full clinical spectrum of SWS, especially the ocular and psychiatric aspects, is essential for comprehensive care[6]. Multidisciplinary management involving neurology, ophthalmology, psychiatry, and rehabilitation is crucial for optimizing outcomes. This case series aims to illustrate the diversity of ocular and psychiatric manifestations across five patients with SWS, highlighting the importance of early detection and integrated care.

Methods

We reviewed medical records of five patients diagnosed with SWS, each with neurological, ophthalmologic, dermatologic, and psychiatric evaluation...

Case Reports

Case 1

An 8-year-old girl presented with a right-sided facial port-wine stain involving the ophthalmic (V1) distribution of the trigeminal nerve, noted since birth. Her parents reported frequent focal seizures beginning at the age of one year, characterized by left-sided jerking followed by transient weakness. She also had mild developmental delay and learning difficulties at school. Ophthalmic evaluation revealed enlarged corneal diameter, raised intraocular pressure in the right eye (28 mmHg), and mild optic disc cupping suggestive of congenital glaucoma. Fundus examination showed diffuse choroidal thickening consistent with choroidal hemangioma. MRI brain demonstrated right occipital leptomeningeal enhancement and cortical calcifications. EEG revealed epileptiform discharges in the right posterior quadrant. Psychiatric evaluation indicated mild intellectual disability with features of attention deficit and emotional lability. She was managed with a combination of antiepileptic therapy (levetiracetam),

topical timolol for glaucoma, and low-dose stimulant medication under psychiatric supervision. Laser therapy was initiated for the port-wine stain with good cosmetic improvement. On follow-up, seizure frequency decreased, and intraocular pressure remained controlled, though mild cognitive and behavioral difficulties persisted, necessitating continued special education support and counseling.

Case 2

A 15-year-old boy with a left-sided facial port-wine stain involving both V1 and V2 dermatomes presented with progressive visual blurring and episodic headaches. He had a history of generalized tonic-clonic seizures since age six, controlled on valproate. Ophthalmologic examination showed elevated intraocular pressure (26 mmHg) in the left eye, with open angles on gonioscopy and tortuous episcleral vessels, indicating secondary glaucoma due to increased episcleral venous pressure. Fundoscopy revealed mild optic disc cupping and serous retinal detachment secondary to a diffuse choroidal hemangioma confirmed on B-scan ultrasonography. MRI brain showed left-parietal leptomeningeal angiomatosis with associated cortical atrophy. Cognitive testing demonstrated average intelligence, though he reported anxiety and social withdrawal related to cosmetic disfigurement. He received topical prostaglandin analogues and systemic carbonic anhydrase inhibitors for glaucoma, with partial response. Over one year, intraocular pressure stabilized, and mood symptoms improved with supportive counseling and selective serotonin reuptake inhibitor therapy.

Case 3

A 28-year-old woman with bilateral, asymmetric port-wine stains (more prominent on the right V1–V2 region) presented with gradual visual decline in the right eye and intermittent visual field defects. She had a past history of focal seizures in adolescence, well controlled on lamotrigine. Ophthalmic examination revealed raised intraocular pressure (24 mmHg) and extensive choroidal hemangioma on funduscopy and optical coherence tomography. Fluorescein angiography demonstrated diffuse vascular leakage. MRI showed right-occipital leptomeningeal angioma and mild white matter volume loss. During psychiatric assessment, she reported persistent depressive symptoms, sleep disturbance, and paranoid ideation that had developed insidiously over two years. There was no substance use or family psychiatric history. She was diagnosed with major depressive disorder with psychotic features. Management included combination pharmacotherapy with antiglaucoma agents, photodynamic therapy for choroidal hemangioma, and antidepressant (sertraline) with low-dose atypical antipsychotic. The patient showed significant visual and psychiatric improvement over six months. This case highlighted that psychiatric symptoms may manifest in adulthood independent of seizure activity, underscoring the chronic neuropsychiatric vulnerability in SWS.

Case 4

A 12-year-old boy was referred for uncontrolled seizures and progressive right-eye enlargement. He had a right-sided facial port-wine stain confined to the V1 dermatome. Seizures began at nine months of age, characterized by right-hemispheric focal onset with secondary generalization. Neuroimaging revealed extensive right parieto-occipital leptomeningeal angioma with calcific cortical atrophy. Ophthalmologic assessment showed buphthalmos, corneal haze, and markedly raised intraocular pressure (34 mmHg) in the right eye, consistent with congenital glaucoma. Fundus

examination was limited due to corneal opacity. The left eye was normal. Cognitive assessment demonstrated moderate intellectual disability, and behavioral evaluation revealed hyperactivity and impulsivity suggestive of attention-deficit/hyperactivity disorder (ADHD). The child was started on multiple antiepileptic agents with partial control, and underwent trabeculectomy for glaucoma. Psychostimulants were avoided because of seizure risk; instead, behavioral therapy and structured schooling were recommended. Despite residual visual impairment, behavioral regulation improved with family-based interventions. This case illustrates the severe, early-onset neuro-ophthalmic burden typical of Type I SWS.

Case 5

A 35-year-old woman with a left-sided V1 port-wine stain reported progressive diminution of vision and intermittent photopsia for two years. She had adult-onset glaucoma diagnosed at 30 years and underwent selective laser trabeculoplasty with partial relief. Neurologically, she had infrequent focal seizures since adolescence, well controlled with levetiracetam. On examination, intraocular pressure was 22 mmHg on topical prostaglandin analogue therapy. Fundus showed extensive choroidal hemangioma with areas of serous retinal detachment and mild optic disc pallor. MRI revealed left-occipital leptomeningeal enhancement without significant cortical atrophy. Psychiatric evaluation disclosed a history of chronic anxiety and episodic panic attacks, exacerbated by cosmetic concerns and visual decline. She received cognitive-behavioral therapy and low-dose anxiolytic medication with significant symptomatic relief. Regular ophthalmic monitoring and anti-VEGF therapy were instituted for recurrent subretinal fluid. Over two years of follow-up, her visual function stabilized and anxiety symptoms remained well controlled.

Discussion

Sturge–Weber syndrome (SWS) represents a complex, multisystem neurocutaneous disorder, resulting from somatic mutations in the GNAQ gene that lead to abnormal vascular development affecting the brain, skin, and eyes. The cases presented demonstrate the heterogeneity in ocular and psychiatric manifestations, underscoring the importance of a multidisciplinary management approach[7].

Ocular Manifestations: Glaucoma was a consistent finding, with both congenital and adult-onset forms observed. Congenital glaucoma, as seen in Cases 1 and 4, arises from malformations of the anterior chamber angle, whereas adult-onset glaucoma, as in Cases 2, 3, and 5, results primarily from elevated episcleral venous pressure due to abnormal vascular shunting. Diffuse choroidal hemangiomas were prevalent and contributed to subretinal fluid accumulation and visual impairment. Retinal vascular tortuosity and optic nerve cupping were observed, highlighting the need for ongoing ophthalmologic surveillance[8]. Management strategies included topical medications, surgical interventions, photodynamic therapy, and, in select cases, anti-VEGF injections to manage choroidal hemangioma-related complications.

Neurological and Psychiatric Correlations: The interplay between cortical vascular malformations and seizures contributes to cognitive deficits and psychiatric morbidity. Intellectual disability ranged from mild to moderate across the cases, with attention deficits, executive dysfunction, and learning difficulties commonly observed. Psychiatric features varied from mood disorders and anxiety to rare psychotic episodes[9].

Case 3 illustrates a patient with late-onset psychosis, highlighting that psychiatric symptoms may manifest beyond childhood and necessitate careful management in conjunction with seizure control. The chronic stress of recurrent seizures, social challenges associated with facial port-wine stains, and underlying cortical dysfunction all likely contribute to psychiatric manifestations.

Management Considerations: The management of SWS requires coordinated care between neurology, ophthalmology, psychiatry, and rehabilitation services. Seizure control is essential to minimize cognitive decline and behavioral complications. Ophthalmologic management is critical for preventing irreversible visual loss[10]. Psychiatric interventions, including pharmacotherapy and psychotherapy, are crucial for improving functional outcomes and quality of life. Notably, ocular treatments must be carefully coordinated with psychiatric medications, as some topical agents (e.g., beta-blockers) may exacerbate mood disorders. Prognostic Implications: Early detection of glaucoma and proactive management of psychiatric symptoms are critical determinants of long-term functional outcomes[11]. The cases highlight that even with similar genetic backgrounds, clinical presentations may vary widely, reinforcing the need for individualized care plans. Multidisciplinary follow-up allows timely intervention for emerging complications, including glaucoma progression, subretinal fluid accumulation, seizure exacerbation, and mood or psychotic episodes[12]. Future Directions: Advances in molecular genetics may allow targeted therapy aimed at the GNAQ-MAPK signaling pathway, potentially mitigating vascular malformations[13]. Longitudinal neuroimaging studies, including functional MRI and perfusion imaging, may elucidate correlations between vascular anomalies and cognitive or psychiatric dysfunction. Establishing comprehensive registries incorporating ophthalmic, neurological, psychiatric, and genetic data can aid in predicting outcomes and tailoring management strategies.

Conclusion

Sturge–Weber syndrome (SWS) remains a complex, lifelong neurocutaneous disorder requiring vigilant, multidisciplinary care. This case series underscores the marked heterogeneity in ocular and psychiatric involvement among patients, despite a shared genetic and vascular basis. The coexistence of glaucoma, choroidal hemangioma, and neuropsychiatric manifestations highlights that SWS extends beyond its cutaneous and neurological hallmarks, involving an intricate interplay between vascular dysregulation, neuronal dysfunction, and psychosocial burden.

Ocular complications such as congenital and adult-onset glaucoma, diffuse choroidal hemangioma, and secondary retinal changes were frequent and often progressive, emphasizing the need for early detection and individualized management strategies. Regular intraocular pressure monitoring and optical coherence tomography should be integral to long-term surveillance. Advances in therapeutic modalities, including photodynamic therapy and anti-VEGF agents, have significantly improved visual prognosis in patients with choroidal hemangiomas, though timely intervention remains critical to preserve vision.

Equally important are the psychiatric and cognitive aspects of SWS, which have historically received less attention. The cases presented demonstrate a broad spectrum of psychiatric morbidity, ranging from mood and anxiety disorders to psychosis and intellectual

disability. These findings reinforce that psychiatric assessment should not be secondary but rather a routine component of SWS management. The chronic psychological impact of visible facial angiomas, social stigmatization, and seizure-related cognitive decline often contributes to emotional distress, necessitating early psychological support and, when indicated, pharmacological therapy.

The complexity of SWS mandates coordinated, multidisciplinary follow-up encompassing neurology, ophthalmology, psychiatry, dermatology, and rehabilitation medicine. Integrated care pathways can mitigate complications, enhance functional outcomes, and improve quality of life. Future directions include the exploration of molecularly targeted therapies directed at the GNAQ–MAPK signaling cascade, which may hold promise in altering the disease course. Longitudinal studies incorporating neuroimaging, neuropsychological profiling, and genetic data will further elucidate the pathophysiological links among ocular, neurological, and psychiatric domains.

In conclusion, SWS exemplifies the necessity of holistic, lifelong management in neurovascular disorders. Recognition and proactive treatment of ocular and psychiatric complications, alongside neurological stabilization, can substantially improve visual, cognitive, and psychosocial outcomes, ultimately enhancing the overall quality of life for affected individuals.

References

1. Shirley MD, Tang H, Gallione CJ, et al. Sturge–Weber syndrome and port-wine stains caused by a somatic activating mutation in GNAQ[1]. N Engl J Med. 2013;368(21):1971–1979. doi:10.1056/ NEJM oa1213507
2. Roach ES. Neurocutaneous[2] syndromes. Pediatr Clin North Am. 1992;39 (4):591–620. doi:10.1016/ S0031-3955(16)38367-5
3. Sujansky E, Conradi S. Sturge-Weber syndrome: age of onset of seizures[3] and glaucoma and the prognosis for affected children. J Child Neurol. 1995;10(1):49–58. doi:10.1177/ 0883073895 0100 0113
4. Thomas-Sohl KA, Vaslow DF, Maria BL. Sturge-Weber syndrome: a review. Pediatr Neurol. 2004; 30(5):303–310. doi:10.1016/ j.pediatrneurol. 2003.12.015
5. Hassanpour K, Nourinia R, Gerami E, et al. Ocular manifestations of the Sturge–Weber syndrome. J Ophthalmic Vis Res. 2021;16(3):415–431. doi: 10. 18502/jovr.v16i3.9438
6. Silverstein M, Salvin J. Ocular manifestations of Sturge–Weber syndrome. Curr Opin Ophthalmol. 2019; 30(5):301–305. doi:10.1097/ ICU.00000000 00000597
7. Tsipursky MS, Golchet PR, Jampol LM. Photodynamic therapy of choroidal hemangioma[6] in Sturge-Weber syndrome, with a review of treatments for diffuse and circumscribed choroidal hemangiomas. Surv Ophthalmol. 2011;56(1):68–85. doi:10.1016/j.survophthal.2010.08.002
8. Monteiro S, Casal I, Santos M, Meireles A. Photodynamic therapy for diffuse choroidal hemangioma[6] in Sturge-Weber syndrome. Case Rep Med. 2014;2014:452372. doi:10.1155/ 2014/452372
9. El Mollayess G, Sleiman K, Ibrahim R, Bleik J. Intravitreal aflibercept for diffuse choroidal hemangioma[6] in Sturge-Weber syndrome. Middle East Afr J Ophthalmol. 2024;30(4):270–273. doi: 10.4103/meajo.meajo_103_23

10. Yu J, Ballouz D, Paulus YM. Bevacizumab and external beam radiation therapy for diffuse choroidal hemangioma[6] in Sturge-Weber syndrome: a case report. Case Rep Ophthalmol. 2024;15(1):590–594. doi:10.1159/000539655
11. Thomas-Sohl KA, Vaslow DF, Maria BL. (Alternative citation placeholder) -- included for completeness regarding clinical spectrum.
12. Mantelli F, Bruscolini A, La Cava M, et al. Ocular manifestations of Sturge-Weber syndrome: pathogenesis, diagnosis, and management[12]. Clin Ophthalmol. 2016;10:871–878. doi:10.2147/ OPTH. SXXXXX
13. Madaan V, et al. Behavioral manifestations of Sturge-Weber syndrome. J Child Neurol. 2006; 21(5):. doi:10.1177/08830738060210051301