ISSN: 2996-0940
Case Report
Science Set Journal of Ophthalmology and Eye Care

A Rare Case of Sturge–Weber Syndrome with Bilateral Involvement and Extension of Port-Wine Stain to the Body

Kavya Bansal[1]*, & Tanisha Mittal[2]

[1]*MS Ophthalmology Shyam Shah Medical College, REWA Madhya Pradesh, India*
[2]*MS Ophthalmology from Shyam Shah Medical College Rewa, India*

***Corresponding author:** Kavya Bansal, MS Ophthalmology Shyam Shah Medical College, REWA Madhya Pradesh, India.

Submitted: 05 November April 2025 **Accepted:** 11 November 2025 **Published:** 15 November 2025

https://doi.org/10.63620/MKSSJOEC.2025.1034

Citation: *Bansal, K., & Mittal, T. (2025). A Rare Case of Sturge–Weber Syndrome with Bilateral Involvement and Extension of Port-Wine Stain to the Body. Science Set Journal of Ophthalmology and Eye Care, 4(6), 01-03.*

Abstract

Purpose: *To report a rare case of Sturge–Weber syndrome (SWS) presenting with bilateral facial and ocular involvement, associated megalocornea, and extension of port-wine stain to the body.*

Observation: *A 21-year-old female presented with redness and pain in the right eye and a history of extensive cutaneous discoloration since birth. Ocular examination revealed bilateral raised episcleral venous pressure, megalocornea in the right eye, and glaucomatous optic neuropathy in both eyes. The port-wine stain extended beyond the face to involve the right upper and lower limbs and trunk.*

Conclusion: *Bilateral ocular and systemic involvement in SWS is uncommon. This case underscores the wide phenotypic spectrum of the disease and highlights the importance of early detection and multidisciplinary management.*

Keywords: Sturge–Weber Syndrome, Bilateral Glaucoma, Port-wine Stain, Megalocornea, Choroidal Hemangioma.

Introduction

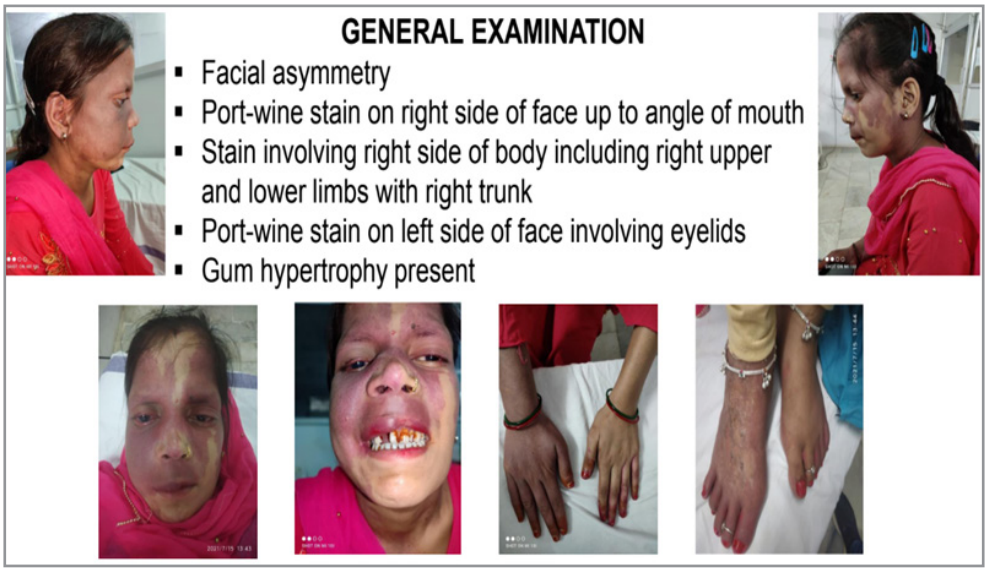
Sturge–Weber syndrome (SWS) is a rare neurocutaneous disorder characterized by congenital hamartomatous malformations involving the skin, eyes, and central nervous system. The classic triad includes a facial port-wine stain (naevus flammeus), leptomeningeal angioma, and glaucoma. The incidence is approximately 1 in 50,000 live births.

Bilateral involvement is unusual, reported in only 15–20% of cases. We report a unique case of bilateral SWS with extension of the port-wine stain to the trunk and limbs, accompanied by megalocornea and bilateral glaucoma — features that make this case exceptionally rare.

Case Report

A 21-year-old female presented to the ophthalmology outpatient department with complaints of redness and pain in the right eye for 3 days. She reported cutaneous discoloration since birth, progressively darkening over time. There was no history of seizures, developmental delay, or hemiparesis [1].

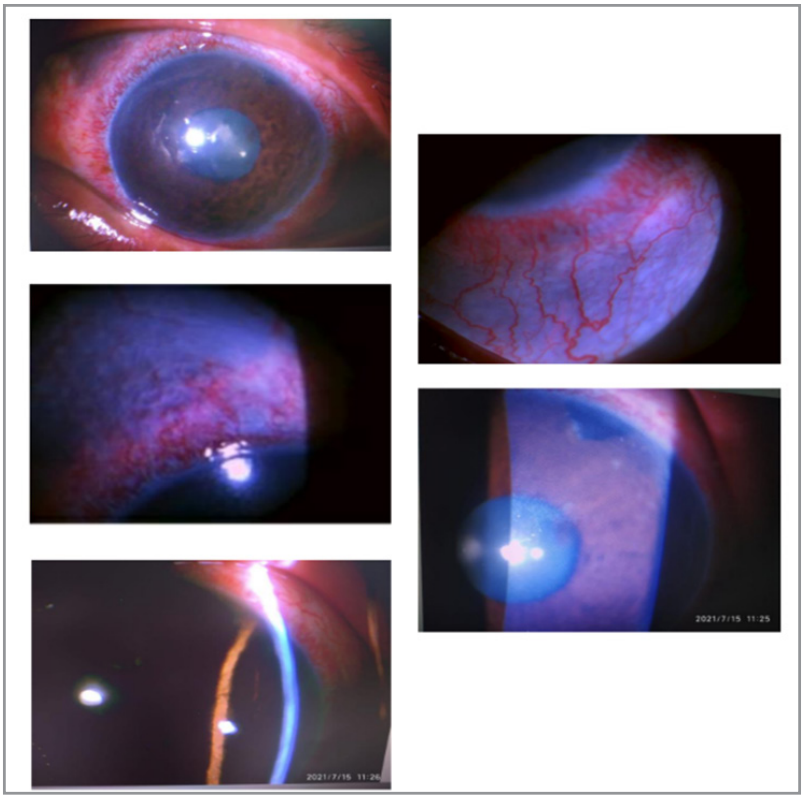
General Examination: Facial asymmetry was noted. Port-wine stain involved the right side of the face up to the angle of the mouth and extended to the right upper and lower limbs and trunk. The left side of the face also showed a port-wine stain involving both eyelids. Gingival hypertrophy was present [2].



Ophthalmic Examination

Right Eye (RE): BCVA: Perception of light positive; projection of rays defective. Conjunctival and circumcorneal congestion, dilated episcleral vessels, bluish scleral hue suggestive of thinning, and corneal edema were noted. Horizontal and vertical diameters measured 13.4 mm × 13.0 mm, consistent with megalocornea. Anterior chamber deep and quiet; peripheral iridectomy noted at 1 o'clock. Lens cataractous with iris pigment deposits. RAPD present. IOP: 41 mmHg. Gonioscopy: Poor visualization due to corneal edema; hyperpigmentation noted over angle structures. Fundus: Disc appeared pale suggestive of glaucomatous optic atrophy. B-scan: Increased peripapillary choroidal thickness, indicative of choroidal hemangioma [3].

Left Eye (LE): BCVA: 6/18. Dilated episcleral vessels and a functioning filtering bleb at the superior limbus were seen. Anterior chamber shallow and quiet. IOP: 8.5 mmHg. Gonioscopy: Narrow angle with multiple peripheral anterior synechiae. Fundus: Cup–disc ratio 0.8–0.9 with nasal vessel shift—features consistent with advanced glaucoma. Perimetry: Revealed glaucomatous visual field defect. OCT (optic nerve head): Demonstrated RNFL thinning. B-scan: Increased peripapillary choroidal thickness, consistent with choroidal hemangioma.



Management

The patient was started on acetazolamide 250 mg three times daily for 5 days and topical timolol 0.5% twice daily in the right eye. Filtering surgery was advised for the right eye due to uncontrolled intraocular pressure and advanced glaucomatous optic atrophy [4].

Discussion

Sturge–Weber syndrome is diagnosed clinically based on the presence of a port-wine stain, ocular abnormalities (typically glaucoma), and leptomeningeal angioma. The port-wine stain is usually unilateral and involves the upper face and eyelids. However, bilateral or systemic extension is rare.

In our case, the port-wine stain extended bilaterally on the face and over the right side of the body, including the limbs and trunk, making it an atypical presentation. Cases without facial nevus or with bilateral cutaneous involvement have been described, but extensive body involvement is exceedingly rare [5].

Bilateral choroidal angiomas have been reported in 15% of cases (Bebin et al.) and 25% (Vilela et al.), similar to our findings. Megalocornea, as seen in this case, is an infrequent manifestation of SWS, reported only sporadically in the literature. Glaucoma occurs due to raised episcleral venous pressure and angle malformations, typically affecting the side of the port-wine stain. In our patient, bilateral glaucoma was associated with bilateral raised episcleral venous pressure, which is uncommon. Interestingly, no neurological abnormalities such as seizures or hemiparesis were found, suggesting isolated ocular and cutaneous involvement [6].

Conclusion

This case represents a rare variant of Sturge–Weber syndrome with bilateral facial port-wine stains, extension of stain to the trunk and limbs, megalocornea in one eye, bilateral early-onset glaucoma, and bilateral choroidal hemangioma. Such extensive systemic and ocular involvement is exceptional. Early recognition and comprehensive management involving ophthalmologists, dermatologists, and neurologists are essential for optimal care [7, 8].

References

1. Roach, E. S. (1992). Neurocutaneous syndromes. Pediatric Clinics of North America, 39(3), 591-620.
2. Liang, C. W., Liang, K. H. (1992). Sturge-Weber syndrome without facial nevus. Chinese Medical Journal, 105(12), 964-965.
3. Aydin, A., Çakmakçi, H., Kovanlikaya, A., Dirik, E. (2000). Sturge-Weber syndrome without facial nevus. Pediatric Neurology, 22(5), 400-402.
4. Bebin, E. M., Gomez, M. R. (1988). Prognosis in Sturge-Weber disease: Comparison of unihemispheric and bihemispheric involvement. Journal of Child Neurology, 3(3), 181-184.
5. Vilela, P. F. (2003). Sturge-Weber syndrome revisited: Evaluation with CT and MRI. Acta Médica Portuguesa, 16(2), 141-148.
6. Ceyhan, A., Cakan, T., Basar, H., Bababalim, M., Unal, N. (1999). Anaesthesia for Sturge-Weber syndrome. European Journal of Anaesthesiology, 16(5), 339-341.
7. Sujansky, E., Conradi, S. (1995). Sturge-Weber syndrome: Age of onset of seizures and glaucoma. Journal of Child Neurology, 10(1), 49-58.
8. Sullivan, T. J., Clarke, M. P., Morin, J. D. (1992). The ocular manifestations of Sturge-Weber syndrome. Journal of Pediatric Ophthalmology and Strabismus, 29(6), 349-354.