Sturge-Weber syndrome with ocular involvement

Síndrome de Sturge-Weber com envolvimento ocular

Oueslati Yassin[1], Selmi Selim[1], Chargui Wael[1], Khammari Houssem Eddine[1], Khallouli Asma[1], Maalej Afef[1]

[1] Department of Ophthalmology, Main Military Training Hospital of Tunis, Tunis, Tunisia.

How to cite:
Yassin O, Selim S, Wael C, Eddine KH, Asma K, Afef M. Sturge-Weber syndrome with ocular involvement. Rev Bras Oftalmol. 2025;84:e0077.

doi:
https://doi.org/10.37039/1982.8551.20250077

Keywords:
Sturge-Weber-syndrome; Neurocutaneous syndrome; Port-wine stain; Glaucoma; Hemangioma

Descritores:
Síndrome de Sturge-Weber; Síndromes neurocutâneas; Mancha vinho do porto; Glaucoma; Hemangioma

ABSTRACT

Sturge-Weber syndrome is a rare sporadic and congenital neurocutaneous disease characterized primarily by the presence of cutaneous and extracutaneous capillary malformations. Our understanding of this syndrome has improved since its first description over the years, particularly with the identification of different clinical presentations and new genetic mutations. Ocular manifestations are frequently observed in this syndrome but often develop insidiously and can threaten visual prognosis. Thus, an appreciation of different ophthalmological manifestations seems crucial. In the same perspective, we report the case of a 12-year-old child who consulted for an ophthalmological examination following a recent discovery of Sturge-Weber syndrome.

RESUMO

A síndrome de Sturge-Weber é uma doença neurocutânea rara, esporádica e congênita, caracterizada principalmente pela presença de malformações capilares cutâneas e extracutâneas. Nossa compreensão dessa síndrome melhorou desde sua primeira descrição ao longo dos anos, especialmente com a identificação de diferentes apresentações clínicas e novas mutações genéticas. As manifestações oculares são frequentemente observadas nessa síndrome, mas geralmente se desenvolvem de forma insidiosa e podem ameaçar o prognóstico visual. Assim, a valorização das diferentes manifestações oftalmológicas parece crucial. Nesse contexto, relatamos o caso de uma criança de 12 anos que procurou atendimento oftalmológico após a descoberta recente da síndrome de Sturge-Weber.

Received on:
December 27, 2024

Accepted on:
September 18, 2025

Corresponding author:
Oueslati Yassin
E-mail: weslatiyassin10@gmail.com

Institution:
Main Military Training Hospital of Tunis, Tunis, Tunisia.

Conflict of interest:
no conflict of interest.

Financial support:
no financial support for this work.

Data Availability Statement:
The datasets generated and/or analyzed during the current study are included in the manuscript.

Edited by:
Editor-in-Chief: Ricardo Augusto Paletta Guedes
Associate Editor: Marcelo Jarczun Kac

INTRODUCTION

Sturge-Weber syndrome (SWS) is a rare congenital non-inherited disorder. It is classically characterized by association of leptomeningeal angiomatosis, facial port-wine birthmark (naevus flammeus), and glaucoma. Ophthalmological presentations are very variable and can affect visual prognosis.

Sturge-Weber syndrome is infrequently associated with oculodermal melanocytosis. This phacomatosis is characterized by excessive development of melanocytes within peripheral ocular skin and diverse ocular structures including uvea, sclera, and iridocorneal angle, giving an appearance of blue-gray and/or brown ocular lesion. Iris involvement is quite characteristic in ocular melanocytosis (OM) with iris mamillations, homogenous protuberances on the iris surface.[1] Patients exhibiting both facial nevus flammeus and OM are classified with phacomatosis pigmentovascularis.[2]

The association of these two phacomatoses, infrequently reported in the literature, exposes patients to earlier and more severe forms of glaucoma by different mechanisms. Therefore, early and effective screening is essential to effectively manage any complications and to preserve vision.

This paper describes the case of a patient, associating type 1 SWS and ocular melanocytosis, to better identify clinical features and possible complications.

Ethical Appreciation Submission Certificate (CAAE) number was 89/2024/CLPP/Hopital Militaire de Tunis.

CASE REPORT

We report the case of a 12-year-old boy, with no relevant medical history, who was referred to our department for a reddish discoloration of the right side of the face.

Inspection found a port wine stain (PWS) of the right hemi-face, extending superiorly from the hairline to the lower edge of the mandible downwards and laterally from the midline of the face to the left ear.

Ophthalmological examination showed buphthalmia of the right eye, congested conjunctival blood vessels, as well as an ocular melanocytosis with extensive slate-gray discoloration of the sclera (Figure 1).

Corrected visual acuity was limited to 2/10 in the right eye and 9/10 in the left eye. Ocular hypertension at 27 mmHg was found in the right eye with hyperpigmentation of the iridocorneal angle. Fundus showed a largely excavated, optic nerve head (Cup/Disc= 0.8) with diffuse chorioretinal atrophy. Examination of the left eye was normal.

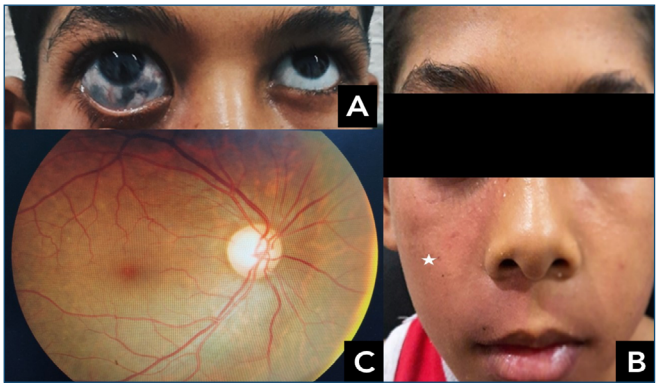

Figure 1. (A) Buphthalmia of the right eye associated with ocular melanocytosis. (B) Port-wine stain on the right side of the face (white star). (C) Fundus examination of the right eye showing pathological papillary excavation with chorioretinal atrophy.

B-mode ocular ultrasound of the right eye found an elongated antero-posterior axis estimated at 32 mm. Sturge-Weber syndrome was suspected. A magnetic resonance imaging scan was performed, and it showed asymmetrical myelination of the right hemisphere as well as a prominent leptomeningeal enhancement in contrast study reflecting pial angiomatosis (Figure 2).

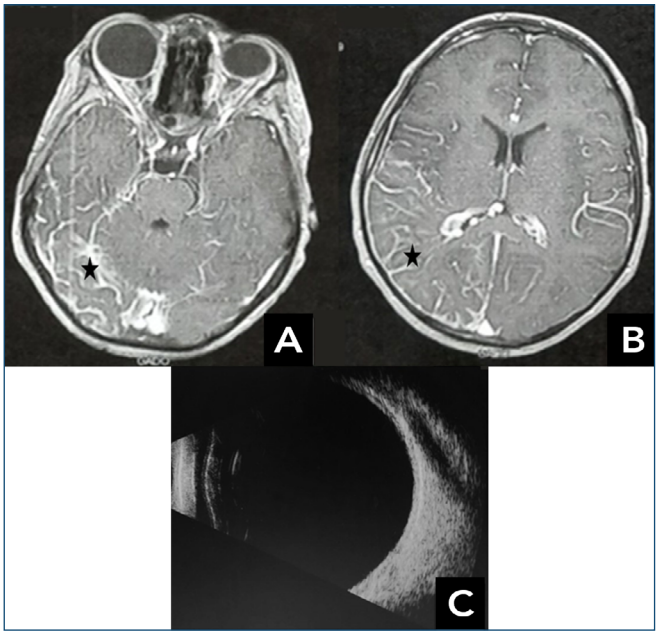

Figure 2. (A, B) Axial T1 weighted magnetic resonance imaging with gadolinium contrast showing contrast enhancement asymmetry with hypersignal on the right side with abnormal venous drainage. (C) Ocular ultrasound of the right eye showing elongation of ocular diameters.

The diagnosis of SWS with ocular involvement (unilateral juvenile glaucoma) was retained. The patient was treated with anti-glaucomatous agents, which failed to normalize intraocular pressure (IOP), so he was referred

to surgical treatment by trabeculectomy, with good post-operative outcomes and control of eye pressure.

DISCUSSION

Sturge-Weber syndrome was first reported by Professor Hilding Bergstrand in 1935 in recognition of the work of William Allen Sturge 1879 and Parkes Weber 1922.[3]

Prevalence of SWS varies from 1/20,000 up to 1/50,000 births and affects both males and females equally without predilection.[4]

Oculodermal melanocytosis is frequently encountered in Asians and Blacks (0.014% to 0.034%) and has been rarely reported in Caucasians.[5] Eyelid skin involvement is noted in one third of the cases; however, the association of eyelid and ocular involvement has been reported in two thirds of the cases.[6]

New genetic studies have improved our knowledge of the pathological process. Two genetic variants GNAQ/GNA11 have been described, causing dysregulation of signal transduction between G protein coupled receptors and downstream effectors, which results in endothelial cell activation, overgrowth of proliferative capillaries, abnormal endothelial cell differentiation, as well as a progressive dilatation of immature venous system.[3,7] Clinical manifestations are variable and have been grouped into three types according to the Roach Scale classification as follows:

- Type 1: both facial and leptomeningeal angiomas; may include glaucoma (classic SWS).
- Type 2: facial angioma alone (no central nervous system involvement); may include glaucoma.
- Type 3: isolated leptomeningeal-brain angioma; usually no glaucoma.[8]

Neurological complications are quite common and affect approximately 70-80% of patients. Leptomeningeal angiomatosis is often unilateral, posterior and on the same side as the facial birthmark. It may include several aspects; mental retardation, seizures or neurological spasms, hemiparesis and behavioral disorder. Seizures are often due to irritation of cerebral cortex by the development of leptomeningeal angioma and may have an early onset in life.[9,10]

Dermatological manifestations are the most characteristic of SWS and often follow a particular anatomical distribution. They manifest as vascular malformations with skin color ranging from pink or red to violaceous patches. Over time, skin lesions tend to become darker with port-wine appearance. They also become increasingly thick and take on a nodular and rough appearance.[11] Risk of associated neurological or ocular damage depends on the extent and location of skin lesions. Extensive and bilateral presentation, hemifacial and forehead localization, median port-wine birthmark, and involvement of more than 50% of contiguous hemi forehead are considered high risk.[12]

Ocular abnormalities are observed in almost half of cases.[13] The two main observed manifestations are glaucoma and choroidal hemangioma (CH). About 30 to 70% of patients develop glaucoma, often ipsilateral to skin lesion or leptomeningeal angiomatosis. Physiopathological mechanisms are multiple and can be explained by an abnormal development of iridocorneal angle, responsible for congenital glaucoma development. Another mechanism involves elevation of episcleral venous pressure associated or not to drainage abnormalities related to outlet resistance of the distal outflow pathway.[13,14] Glaucoma is often refractory, resistant to medical treatment and frequently requiring surgical treatment.

Medical treatment is frequently employed for a temporarily regulation of IOP, either as a transient measure prior to surgery or as an adjunct to postoperative management.[15] Both topical beta-adrenergic antagonists and carbonic anhydrase inhibitors, administered as monotherapy or in combination, represent the most widely used and efficacious agents.[6] Glaucoma associated with iridocorneal angle pigment dispersion treatment relies on selective laser trabeculoplasty.[16]

Surgical approaches often remain the definitive management strategy. Goniotomy or trabeculotomy may be performed, in the absence of angular synechiae, to reduce IOP by addressing chamber angle abnormalities. Alternatively, filtering surgery, such as trabeculectomy or drainage valve implantation, are employed to create a secondary drainage pathway.[6] Kaushik et al. reported a 75% success rate following Ahmed valve implantation, with a significant reduction in IOP and number of pharmacological needed agents.[17]

Choroidal hemangioma is observed in 40% to 50% of cases. Diagnosis is evoked on fundus examination with a characteristic appearance of enlarged vessels and vascular channels. These abnormalities may be responsible for several anomalies, namely visual acuity reduction, scotoma, or even retinal detachment.[14,18]

Treatment of CH is attempted only when visual function is threatened by complications such as exudative retinal detachment.[6] Therapeutic strategy depends on nature and location of the hemangioma. Diffuse CH are

treated by stereotactic radiotherapy, external beam radiotherapy and proton beam radiotherapy. Argon laser is of interest in the treatment of circumscribed forms in association with therapeutic means mentioned above.[19] Photodynamic therapy with verteporfin is a new non-invasive therapeutic modality that allows shrinkage of tumor size and reduces its diffusion.[20] It represents the treatment of choice in circumscribed hemangioma but also proves encouraging results in diffuse forms.[21] Other ocular abnormalities have been reported with varying frequency, such as iris heterochromia, iris mammillation, vascular occlusion.[14]

Diagnosis and treatment of the various complications associated with SWS involves lifelong surveillance from birth, followed by regular ophthalmological follow-up.[22] Papillary optical coherence tomography (OCT) facilitates early detection of glaucomatous deficits even in nascent stages, while visual field testing is more reliable in adult populations.[23] Ultrasound biomicroscopy provides high-resolution study of the iridocorneal angle. Similarly, regular follow-up combining fundus examination, ultrasound biomicroscopy and OCT of the posterior pole is crucial for uveal melanoma screening, particularly in the presence of iris mammillations.[24]

In conclusion, Sturge-Weber syndrome is a neurocutaneous disorder with variable clinical features and can be associated with severe complications threatening functional as well as vital prognosis. From birth, lifelong surveillance, associated with meticulous ophthalmological follow-up, allows a proactive approach that facilitates prompt therapeutic intervention and avoids the risk of irreversible visual impairment, by emphasizing the value of consistent monitoring and leveraging advanced diagnostic

AUTHORS' CONTRIBUTION

Oueslati Yassin and Selim Selmi contributed to the conception writing and critical review of the manuscript's content. Chargui Wael and Khammari Houssem Eddine contributed to writing and illustration preparation. Khallouli Asma and Maalej Afef contributed to critical review of the manuscript. All authors approved the final version of the manuscript and are responsible for all aspects of the manuscript, including ensuring its accuracy and integrity.

REFERENCES

1. Gilliam AC, Ragge NK, Perez MI, Bolognia JL. Phakomatosis pigmentovascularis type IIb with iris mammillations. Arch Dermatol. 1993;129(3):340-2.
2. Plateroti AM, Plateroti R, Mollo R, Librando A, Contestabile MT, Fenicia V. Sturge-Weber Syndrome Associated with Monolateral Ocular Melanocytosis, Iris Mammillations, and Diffuse Choroidal Haemangioma. Case Rep Ophthalmol. 2017;8(2):375-84.
3. Sudarsanam A, Ardern-Holmes SL. Sturge-Weber syndrome: from the past to the present. Eur J Paediatr Neurol. 2014;18(3):257-66.
4. Singh AK, Keenaghan M. Sturge-Weber Syndrome. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2024 [cité 22 mars 2024]. Disponible sur: http://www.ncbi.nlm.nih.gov/books/NBK459163/
5. Chan HH, Kono T. Nevus of Ota: Clinical aspects and management. Skinmed. 2003;2(2):89-98.
6. Abdolrahimzadeh S, Pugi DM, De Paula A, Scuderi G. Ocular manifestations in phakomatosis pigmentovascularis: Current concepts on pathogenesis, diagnosis, and management. Survey of Ophthalmology. 2021;66(3):482-92.
7. Dutkiewicz AS, Ezzedine K, Mazereeuw-Hautier J, Lacour JP, Barbarot S, Vabres P, et al. A prospective study of risk for Sturge-Weber syndrome in children with upper facial port-wine stain. J Am Acad Dermatol. 2015;72(3):473-80.
8. Roach ES. Neurocutaneous syndromes. Pediatr Clin North Am. 1992;39(4):591-620. doi: 10.1016/s0031-3955(16)38367-5
9. Comi AM. Presentation, diagnosis, pathophysiology, and treatment of the neurological features of Sturge-Weber syndrome. Neurologist. 2011;17(4):179-84.
10. Pascual-Castroviejo I, Pascual-Pascual SI, Velazquez-Fragua R, Viaño J. Sturge-Weber syndrome: study of 55 patients. Can J Neurol Sci. 2008;35(3):301-7.
11. Passeron T, Salhi A, Mazer JM, Lavogiez C, Mazereeuw-Hautier J, Galliot C, et al. Prognosis and response to laser treatment of early-onset hypertrophic port-wine stains (PWS). J Am Acad Dermatol. 2016;75(1):64-8.
12. Sabeti S, Ball KL, Bhattacharya SK, Bitrian E, Blieden LS, Brandt JD, et al. Consensus Statement for the Management and Treatment of Sturge-Weber Syndrome: Neurology, Neuroimaging, and Ophthalmology Recommendations. Pediatr Neurol. 2021;121:59-66.
13. Higueros E, Roe E, Granell E, Baselga E. Sturge-Weber Syndrome: A Review. Actas Dermosifiliogr. 2017;108(5):407-17.
14. Hassanpour K, Nourinia R, Gerami E, Mahmoudi G, Esfandiari H. Ocular Manifestations of the Sturge–Weber Syndrome. J Ophthalmic Vis Res. 2021;16(3):415-31.
15. Abdolrahimzadeh S, Fameli V, Mollo R, Contestabile MT, Perdicchi A, Recupero SM. Rare diseases leading to childhood glaucoma: epidemiology, pathophysiogenesis, and management. Biomed Res Int. 2015;2015:781294.
16. Scuderi G, Contestabile MT, Scuderi L, Librando A, Fenicia V, Rahimi S. Pigment dispersion syndrome and pigmentary glaucoma: a review and update. Int Ophthalmol. 2019;39(7):1651-62.
17. Kaushik J, Parihar JK, Jain VK, Mathur V. Ahmed valve implantation in childhood glaucoma associated with Sturge-Weber syndrome: our experience. Eye (Lond). 2019;33(3):464-8.
18. Formisano M, Abdolrahimzadeh B, Mollo R, Bruni P, Malagola R, Abdolrahimzadeh S. Bilateral diffuse choroidal hemangioma in Sturge-Weber syndrome: A case report highlighting the role of multimodal imaging and a brief review of the literature. J Curr Ophthalmol. 2018;31(2):242-9.
19. Sánchez-Espino LF, Ivars M, Antoñanzas J, Baselga E. Sturge-Weber Syndrome: A review of pathophysiology, genetics, clinical features, and current management approache. Appl Clin Genet. 2023;16:63-81. Erratum in: Appl Clin Genet. 2024;17:131-132.
20. Tsipursky MS, Golchet PR, Jampol LM. Photodynamic therapy of choroidal hemangioma in sturge-weber syndrome, with a review of treatments for diffuse and circumscribed choroidal hemangiomas. Surv Ophthalmol. 2011;56(1):68-85.
21. Anand R. Photodynamic therapy for diffuse choroidal hemangioma associated with Sturge-Weber syndrome. Am J Ophthalmol. 2003;136(4):758-60.
22. Thavikulwat AT, Edward DP, AlDarrab A, Vajaranant TS. Pathophysiology and management of glaucoma associated with phakomatoses. J Neurosci Res. 2019;97(1):57-69.

23. Perdicchi A, de Paula A, Sordi E, Scuderi G. Cluster analysis of computerized visual field and optical coherence tomography-ganglion cell complex defects in high intraocular pressure patients or early stage glaucoma. Eur J Ophthalmol. 2020;30(3):475-9.

24. Gündüz K, Shields CL, Shields JA, Eagle RC, Singh AD. Iris mammillations as the only sign of ocular melanocytosis in a child with choroidal melanoma. Arch Ophthalmol. 2000;118(5):716-7.