Unilateral port wine stain on the face: A case report and review

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ABSTRACT

Introduction: Sturge-Weber syndrome (SWS) is a sporadic, progressive, congenital condition that occurs due to hamartomatous malformation and is usually referred to as a "port wine stain." It is characterized by trisymptomatic forms that include facial port wine stain, glaucoma, and leptomeningeal calcifications. **Case description:** A 42-year-old female patient presented with a chief complaint of missing teeth in the mandibular anterior region that had recently exfoliated following mobility. A unilateral port wine stain noted on the left side of her face appeared to follow the left maxillary division of the trigeminal nerve with minimal intraoral signs. The patient claimed it had been present since birth. **Discussion:** The origin, pathophysiology, clinical presentation, differential diagnosis, potential therapies, and prognosis of SWS are discussed. **Conclusion:** A multidisciplinary approach to individuals with SWS is required for the successful treatment of these patients.

RÉSUMÉ

Introduction : Le syndrome de Sturge-Weber (SSW) est une affection congénitale, sporadique et progressive, résultant d'une malformation hamartomateuse et généralement désignée sous le terme de « tache de vin ». Il se caractérise par une forme trisymptomatique associant une tache de vin du visage, un glaucome et des calcifications leptoméningées. **Description du cas :** Une patiente de 42 ans s'est présentée avec comme plainte principale l'absence de dents dans la région antérieure mandibulaire, récemment exfoliées à la suite d'une mobilité dentaire. Une tache de vin unilatérale, observée sur le côté gauche de son visage, semblait suivre la division maxillaire gauche du nerf trijumeau, avec des signes intra-oraux minimes. La patiente a déclaré que cette lésion était présente depuis la naissance. **Discussion :** L'origine, la physiopathologie, la présentation clinique, le diagnostic différentiel, les options thérapeutiques potentielles ainsi que le pronostic du syndrome de Sturge-Weber sont discutés. **Conclusion :** Une prise en charge multidisciplinaire des personnes ayant le syndrome de Sturge-Weber est nécessaire pour assurer le succès du traitement de ces patients.

Keywords: congenital; face; hemangioma; port wine stain; Sturge-Weber syndrome; vascular malformations
CDHA Research Agenda category: risk assessment and management

INTRODUCTION

Sturge-Weber syndrome (SWS), also referred to as an encephalofacial angiomatosis or encephalotrigeminal angiomatosis, is a rare disorder classified as a mesodermal phakomatosis. It is commonly referred to as either a port wine stain or a nevus flammeus.[1] It is a congenital, sporadic, neurocutaneous disorder characterized by the presence of a facial port wine stain, glaucoma, and leptomeningeal angiomatosis.[2] It is also recognized as a fourth phacomatosis, because it is characterized by nevus flammeus of the face and angioma of the meninges.[3] There are 3 types of SWS: Type 1 involves a facial port wine stain and leptomeningeal angioma with possible ocular abnormalities such as glaucoma; Type 2 involves a facial port wine stain (with possible glaucoma); and Type 3 exclusively involves a leptomeningeal angioma.[4] Males and females are equally affected, with no racial predilection.[5] Etiology is uncertain, but it is thought to be caused by persistence of vascular plexus around the cephalic portion of the neural tube that develops during the sixth week of intrauterine life and regresses during the ninth week of intrauterine life due to mutation of a gene (GNAQ).[5] It is recognized as the third most common neurocutaneous syndrome.[2]

SWS is observed in areas supplied by the trigeminal nerve (fifth cranial nerve or C5), most primarily marked by the facial port wine stain in the V2 distribution of C5 that covers the facial region.[6] Rudolf Schirmer was the first to identify this illness in 1860. William Allen Sturge then gave a detailed description of the syndrome in 1879, while Frederick Parker Weber later showed cerebral

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calcifications in 1922. In 1935, Swedish physician Hilding Bergstrand named the condition Sturge-Weber syndrome in recognition of its original describers.[5] A case report of a patient with facial port wine stains more indicative of Type 2 SWS is presented here.

CASE DESCRIPTION

A 42-year-old female patient reported to the Department of Oral Medicine and Radiology at the Sree Mookambika Institute of Dental Sciences, India, with a chief complaint of missing teeth in the mandibular anterior region within the past week. She disclosed a history of tooth exfoliation following mobility. She had lost upper right and left first molars and 2 lower anterior teeth. According to the patient's medical history, she indicated taking medicine for the last 1.5 years for diabetes mellitus, and for hypercholesterolemia for the last 3 months. She also mentioned the purplish discolouration on the left side of her face, reporting it had existed from birth. Initially, she indicated it was small but grew to its current size and became darker with age. No prior history of any neurological problems, such as headaches or seizures, were reported. Family history was non-contributory.

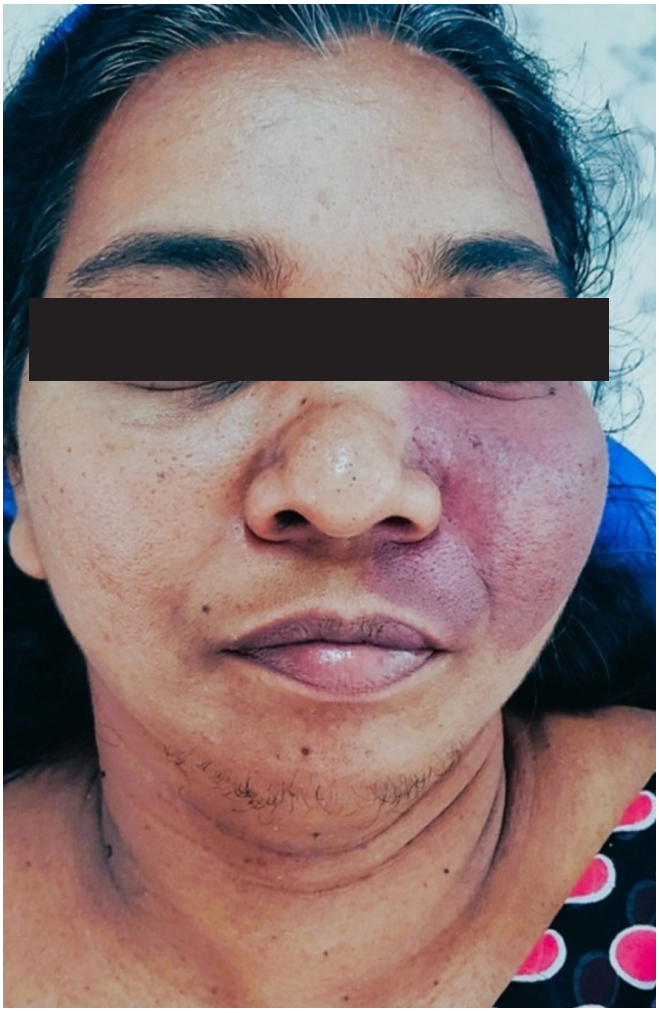

Figure 1. Port wine stain on the left midface region along the distribution of maxillary branch of trigeminal nerve

Upon extraoral examination, a unilateral distribution of a port wine stain with purplish discolouration was noted on the left side of the face. It began at the lower left eyelid and spread to the lateral aspect of the nose, philtrum, and left side of the upper lip 5 mm above the vermillion border and left cheek. The port wine stain did not cross the midline of the face; it exclusively affected the left side of the face. There was no involvement of the lower lip or lower jaw (Figure 1).

The oral hygiene of the patient was poor with extensive amounts of debris and calculus. The gingiva was inflamed, with bleeding on probing, and the radiograph revealed generalized bone loss indicative of chronic generalized periodontitis. No obvious gingival hyperplasia was evident. There was an erythematous zone on the left buccal mucosa extending from the first premolar region anteriorly to the second molar region posteriorly, as well as up to the vestibular level superiorly and along the occlusal plane inferiorly, which represented the extension of the lesion intraorally (Figure 2).

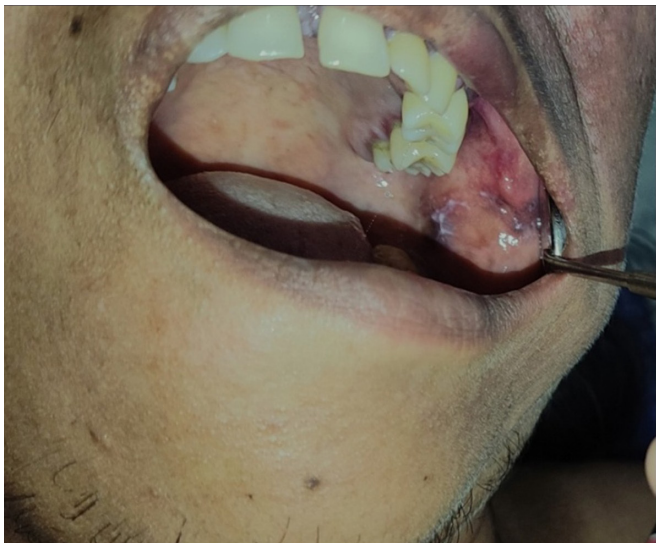

Figure 2. Intraoral extension of the port wine stain on the left buccal mucosa

Hemangioma may be regarded as a potential differential diagnosis for these findings given that the lesion is congenital. However, diascopy should yield a positive result for hemangioma, whereas in this case it was negative, which supports the diagnosis of SWS.[7] Another differential diagnosis for port wine stains is nevus simplex or salmon patches. Salmon patches are cutaneous venous malformations that are typically found on the forehead (also called an angel's kiss), nape (also called stork bite marks), and upper eyelids. Most salmon patches disappear between the ages of 1 and 2.[8]

Based on these clinical features, despite the lack of

neurological symptoms such as mental impairment, seizures, headaches, fever, vomiting, head injuries, stroke-like episodes, dizziness or impaired eyesight that are typical signs of SWS, the patient was diagnosed with SWS.

As the patient is not in the high-risk category, no imaging modalities (such as magnetic resonance imaging) were carried out to rule out cranial involvement. Clinical findings were contributory. Sanchez Espino et al.[2] stated that asymptomatic children above 2 years of age may not need central nervous system (CNS) imaging (Table 1).

Table 1. Patients at high risk of CNS abnormalities based on the location and extension of the port wine stain

Criteria
Extensive bilateral involvement
Hemifacial and forehead involvement (± upper eyelid)
Median port wine birthmark[a]
>50% of contiguous hemi forehead involvement.

[a]Linear extension from the medial forehead, glabella, and base of the nose[2]

However, an orthopantomograph was taken for the patient, which showed chronic generalized periodontitis, missing upper right and left first molars and 2 lower front teeth (Figure 3).

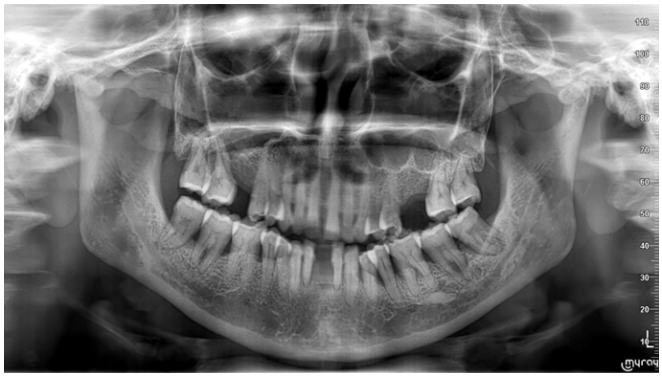

Figure 3. Orthopantomograph showing generalized periodontitis and missing upper right and left first molars and lower front teeth

The patient's oral plaque control regimen was established as part of the treatment. Instructions were given on oral hygiene maintenance and the use of chlorhexidine mouthrinse in addition to oral prophylaxis, followed by prosthetic rehabilitation of the missing teeth.

DISCUSSION

SWS is characterized by a congenital hamartomatous malformation that usually affects the skin, eyes, and CNS, and presents as a combination of venous angioma of the face, eye, and leptomeninges.[9] Apart from port wine stain, another notable clinical symptom of SWS is leptomeningeal angiomatosis, which may result in contralateral enlargement, cerebral calcification, epileptic convulsions, and cognitive impairment. Typically, leptomeningeal angiomas are aberrant blood vessel growth inside the 2 thin tissue layers covering the brain and spinal cord.[5] Leptomeningeal angiomas are typically found unilaterally, particularly in the parietal and occipital areas. In addition, by the time children with SWS are 3 years old, over 70% have partial seizures, and between 50% and 75% show signs of intellectual disability or developmental delay.[1] The less common form, which only involves leptomeningeal angioma, has been defined as Type 3 SWS. It is the rarest subtype, and it is difficult to diagnose as it does not involve skin abnormalities.[10]

SWS occurs in 1 in 50,000 people.[11] This syndrome occurs unilaterally most often but in some cases it can also occur bilaterally.[1] The nevus of the face present from birth becomes darker with age.[3] This syndrome occurs due to post-zygomatic, somatic mosaic mutations in the GNAQ gene, which is located in the long arm of chromosome 9.[1,8] In childhood, SWS angiomas are usually faint, pink macules that tend to darken to red purple; they may be isolated with a well-delineated border, or may be very diffuse.[12]

Due to the patient's clinical presentation and lack of neurological involvement, this case was classified as Type 2 SWS on the Roach scale. The Roach scale helped in the classification of the condition.[13]

- Type 1: Both facial and leptomeningeal angiomas, may have glaucoma
- Type 2: Facial angiomas alone, may have glaucoma
- Type 3: Isolated leptomeningeal angioma, usually no glaucoma

Unilateral port wine stain was the only clinical presentation in the present case. SWS can be categorized as incomplete when it involves only one area without involving the others, and it is said to be complete when it involves both CNS and facial angiomas.[1]

Interestingly, some cases of facial port wine stain with gingival hypertrophy have been reported to occur due to an angiomatosis growth superimposed with poor oral hygiene.[14] In some cases, there may be macroglossia and maxillary bone hypertrophy that lead to malocclusion and a dysmorphic facial appearance.[5] These changes were not evident in the present case.

If clinical signs are not clearly evident, it is essential to assess the extent of intracranial involvement through imaging.[1] Sometimes on computed tomography, MRI or other radiographic images, the presence of gyriform

calcifications, often referred to as a "tram track sign" or "railroad-track sign" have been observed.[1,5,15]

In the present case, treatment outcomes were favourable. However, if a patient with this condition requires any invasive procedures in the maxillofacial region, such as dental extractions, gum surgery or dental implants, they must be done in a hospital setting, as achieving hemostasis can be a major problem. Hemostatic medicines such as topical bovine thrombin, postoperative splints, injection of sclerosing solutions, percutaneous transcatheter vascular embolization with gelfoam or polyvinyl alcohol, and blood transfusion availability can all be used to reduce the risk of bleeding.

Several treatment options are available for individuals with SWS. For regression of well-localized small lesions, high doses of hydrocortisone can be given orally.[2] For lightening of the port wine birthmark, flashlamp-pumped pulsed dye laser (PDL) have been used, targeting only the port wine birthmark without affecting the epidermis and dermis.[6] Sirolimus can be given to improve the prognosis of vascular tumours, venous and lymphatic malformations, and it can also control epilepsy. Topical rapamycin in combination with PDL is more effective in the pediatric population. Rapamycin is given for reduction of soft-tissue overgrowth, attenuation of capillary discolouration, and reduction of ocular pressure. In infants with bilateral facial and extensive leptomeningeal involvement, rapamycin in combination with aspirin is given prophylactically. Cryosurgery can be used to correct lip and soft tissue deformities.[2] Education about plaque and calculus control measures is imperative for maintaining good oral hygiene.

CONCLUSION

Patients with Sturge-Weber syndrome must be informed, along with their parents, that although there is no known cure for this illness, there are options to manage the neurological and ocular symptoms as well as new treatments that can minimize the appearance of the stain and soft-tissue overgrowth. Maintaining proper oral hygiene, ensuring regular gingival and periodontal debridement, and booking regular oral health care appointments are paramount to the prevention of complications from oral lesions and enhancement of the quality of life of patients with Sturge-Weber syndrome.

CONFLICTS OF INTEREST

The authors have declared no conflicts of interest.

INFORMED CONSENT

Written informed consent was obtained from the patient for the publication of images related to her case in this short communication.

REFERENCES

1. Sherwani OA, Patra PC, Ahmad SA, Hasan S. Sturge-Weber syndrome: a report of a rare case. *Cureus.* 2023;15(12):e51110. doi.org/10.7759/cureus.51110.
2. Sánchez-Espino LF, Ivars M, Antoñanzas J, Baselga E. Sturge-Weber syndrome: a review of pathophysiology, genetics, clinical features, and current management approach. *Appl Clin Genet.* 2023;16:63–81. doi.org/10.2147/tacg.s363685. Erratum in: *Appl Clin Genet.* 2024;17:131–32. doi: 10.2147/TACG.S487419
3. Parisi L, Di Filippo T, La Grutta S, Lo Baido R, Epifanio MS, Esposito M, et al. Sturge-Weber syndrome: a report of 14 cases. *Ment Illn.* 2013;5(1):e7. Available from: pubmed.ncbi.nlm.nih.gov/25478131
4. Koenraads Y, van Egmond-Ebbeling MB, de Boer JH, Imhof SM, Braun KP, Porro GL; SWS study group. Visual outcome in Sturge-Weber syndrome: a systematic review and Dutch multicentre cohort. *Acta Ophthalmol.* 2016;94(7):638–45. Available from: pubmed.ncbi.nlm.nih.gov/27238857
5. Babaji P, Bansal A, Choudhury GK, Nayak R, Kodangala Prabhakar A, Suratkal N, et al. Sturge-Weber syndrome with osteohypertrophy of maxilla. *Case Rep Pediatr.* 2013;2013:964596. Available from: pmc.ncbi.nlm.nih.gov/articles/PMC3681312
6. Bachur CD, Comi AM. Sturge-Weber syndrome. *Curr Treat Options Neurol.* 2013;15(5):607–17. doi.org/10.1007/s11940-013-0253-6
7. R IJM, Arumugam Venkatachalam Sargurunathan E, Gowda Venkatesha RR, Rajaram Mohan K, Fenn SM. Port-wine stains and intraoral hemangiomas: a case series. *Cureus.* 2024;16(6):e63532. doi.org/10.7759/cureus.63532
8. Leung AK, Barankin B, Hon KL. Persistent salmon patch on the forehead and glabellum in a Chinese adult. *Case Rep Med.* 2014;2014:139174. Available from: pmc.ncbi.nlm.nih.gov/articles/PMC4053299.
9. Raval DM, Rathod VM, Patel AB, Sharma B, Lukhi PD. Sturge-Weber syndrome: a rare case report. *Cureus.* 2022;14(9):e28786. doi.org/10.7759/cureus.28786
10. Tekin HG, Gökben S, Yılmaz S, Tekgül H, Serdaroğlu G. Sturge-Weber syndrome type III. *J Pediatr Res.* 2018;5(2):103–105. doi.org/10.4274/jpr.44265
11. Neerupakam M, Reddy PS, Babu BA, Krishna GV. Sturge Weber syndrome: a case study. *J Clin Diagn Res.* 2017;11(5):ZD12-ZD14. doi.org/10.7860/jcdr/2017/25593.9891
12. Maruani A. Syndrome de Sturge-Weber [Sturge-Weber syndrome]. *Presse Med.* 2010;39(4):482–86. Available from: pubmed.ncbi.nlm.nih.gov/20219318
13. Gill NC, Bhaskar N. Sturge–Weber syndrome: a case report. *Contemp Clin Dent.* 2010;1(3):183–85. doi.org/10.4103/0976-237x.72789
14. Manivannan N, Gokulanathan S, Ahathya RS, Gubernath, Daniel R, Shanmugasundaram. Sturge-Weber syndrome. *J Pharm Bioallied Sci.* 2012;4(Suppl 2):S349–S352. url: https://doi.org/10.4103/0975-7406.100304
15. Timilsina S, Kunwor B, Thapa Chhetri S, Nepal S, Sedhai K. Sturge-Weber syndrome: a case report. *J Nepal Med Assoc.* 2023;61(267):890–92. doi.org/10.31729/jnma.8344