Unilateral portwine 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 which occurs due to hamartomatous malformation and is usually referred to as a "portwine stain". It is characterized by tri-symptomatic forms that include facial port-wine stain, glaucoma and leptomeningeal calcifications. **Description of case**: This is a case report of a 42-year-old female patient who presented with a chief complaint of missing teeth in the mandibular anterior region that had recently exfoliated following mobility. A unilateral port wine stain was noted on the left side of her face that 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 Sturge-Weber Syndrome are discussed. **Conclusion**: The necessity of a multidisciplinary approach to individuals with SWS is required for the successful treatment of these 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 is a rare disorder classified as a mesodermal phakomatosis which is also referred as an encephalo-facial angiomatosis or encephalo-trigeminal angiomatosis. It is also commonly referred to as either a portwine 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 known as forthfacomatosis, because it is characterized by nevus flammeus of the face and angioma of the meninges.[3] There are three types of SWS, where type 1 includes 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 6th week of intrauterine life and regressess during the 9th 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 (5th 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] Schirmer was the first to identify this illness in 1860. Sturge then gave a detailed description of the syndrome in 1879, while Frederick Parker Weber later showed cerebral calcifications in 1922. In 1935, Swedish physician Hilding Bergstrand named the condition Sturge-Weber syndrome in remembrance 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 REPORT

A 42 year old female patient reported to the Department of Oral medicine and Radiology 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 three months. She also mentioned the purplish discoloration 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.

Upon extraoral examination, a unilateral distribution of a port wine stain with purplish discoloration 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, 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; exclusively affected only 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.

Gingiva was inflamed with bleeding on probing and the radiograph revealed generalized bone loss that is 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 picturizes the extension of the lesion intraorally. [Figure 2]

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 our case it was negative, which supports the diagnosis of SWS. [7]Another differential diagnosis for portwine stains is nevus simplex or salmon patches. Salmon patches, sometimes called nevus simplex, are cutaneous venous malformations that are typically found on the forehead (also called an angel's kiss), nape of the neck (also called stork bite marks), and upper eyelids. Between the ages of one and two, most salmon spots disappear.[8]

Based on these clinical features despite 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 our patient is not in the high risk category, no imaging modalities (like MRI) were carried out to rule out the cranial involvement. Clinical findings itself were contributory. Sanchez Espino et al. stated that asymptomatic children above 2 years of age may not need CNS imaging.[2]

High-Risk Patients for Central Nervous System (CNS) Abnormalities Based on the Location and Extension of the Port-Wine Stain.

- Extensive bilateral involvement.
- Hemifacial and forehead involvement (± upper eyelid).
- Median port-wine birthmark.*
- >50% of contiguous hemi forehead involvement.

Note: *Linear extension from the medial forehead, glabella, and base of the nose.[2]

However OPG (Orthopantomograph) was taken for the patient that showed chronic generalized periodontitis, missing upper right and left first molars and 2 lower front teeth. [Figure 3]

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 mouth rinse 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 central nervous system (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 two thin tissue layers covering the brain and spinal cord.[5] Leptomeningeal angiomas are typically found unilaterally, particularly in the parietal and occipital areas. Additional by the time they are three years old, over 70% of kids with SWS have partial seizures, and between 50% and 75% of them show signs of mental retardation or developmental delay.[1]The less common form, which can be difficult to diagnose and only involves leptomeningeal angioma, has been defined as Type III SWS. Type III Sturge-Weber syndrome 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 population.[11] Mostly this syndrome occurs unilaterally but in some cases it can also occur bilaterally.[1] The nevus of the face present from the birth becomes darker with increase in age.[3] This syndrome occurs due to a post zygomatic, somatic mosaic mutations in the GNAQ gene which is located in the long arm of chromosome 9. [(1,8)] In childhood, SWS are usually faint, pink macules, that tend to darken to red purple; it may be isolated with a well delineated border, or may be very diffuse.[12]

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

Type I. Both facial and leptomeningeal angiomas, may have glaucoma.

Type II. Facial angiomas alone, may have glaucoma.

Type III. 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 portwine 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 our case.

If clinical signs are not clearly evident, it is essential to assess the extent of intracranial involvement through imaging.[1] Sometimes on CT, MRI or other radiographic images, presence of gyriform calcifications, often referred to as a “tramtrack sign” or “railroad-track sign have been observed. [(1,5,15)]

In the present case, treatment outcomes were favourable. But if the patient requires any invasive procedures in the maxillofacial region like dental extractions, gum surgery and dental implants, the same can be done only in a hospital setting, as achieving hemostasis can be a major problem. Haemostatic 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-localised small lesions, high doses of hydrocortisone can be given orally.[2] For lightening of the port-wine birthmark, flashlamp-pumped PDL(pulsed dye laser) have been used, that targets 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(pulsed dye laser) is more effective in the pediatric population. Rapamycin is given for reduction of soft-tissue overgrowth, attenuation of capillary discoloration 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] Needless to say, education about plaque and calculus control measures is imperative for maintaining good oral hygiene.

CONCLUSION

For patients with Sturge-Weber Syndrome, it is important to inform them and their parents that although there is no known cure for this illness, there are not only options to manage the neurological and ocular symptoms but there are also new treatments that can minimize the appearance of the stain and soft-tissue overgrowth. Maintaining proper oral hygiene, regular gingival and periodontal debridement and regular dental checkups is paramount to the prevention of complications from oral lesions and enhancement of the quality of life for patients with Sturge-Weber syndrome.

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FIGURES:

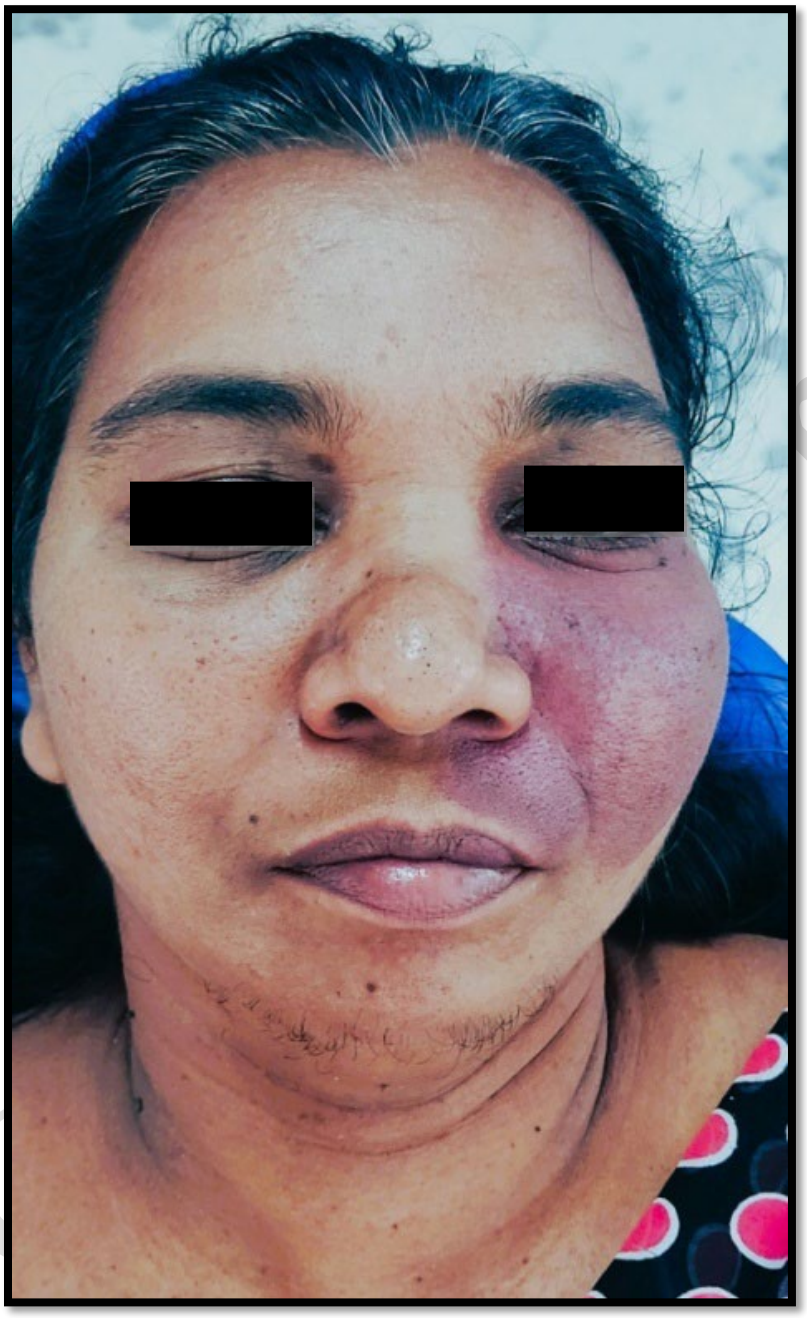

Figure 1: The port wine stain on the left midface region along the distribution of maxillary branch of trigeminal nerve.

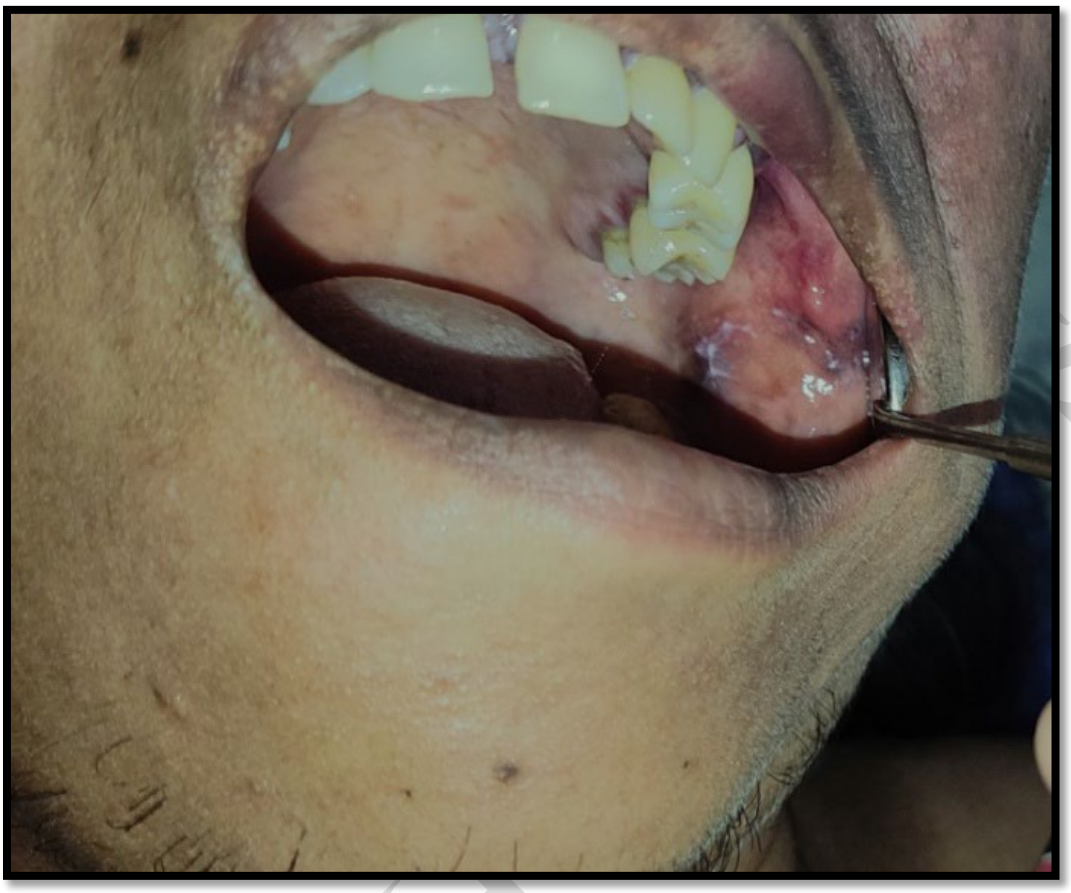

Figure 2: Intraoral extension of the Port wine stain on the left buccal mucosa.

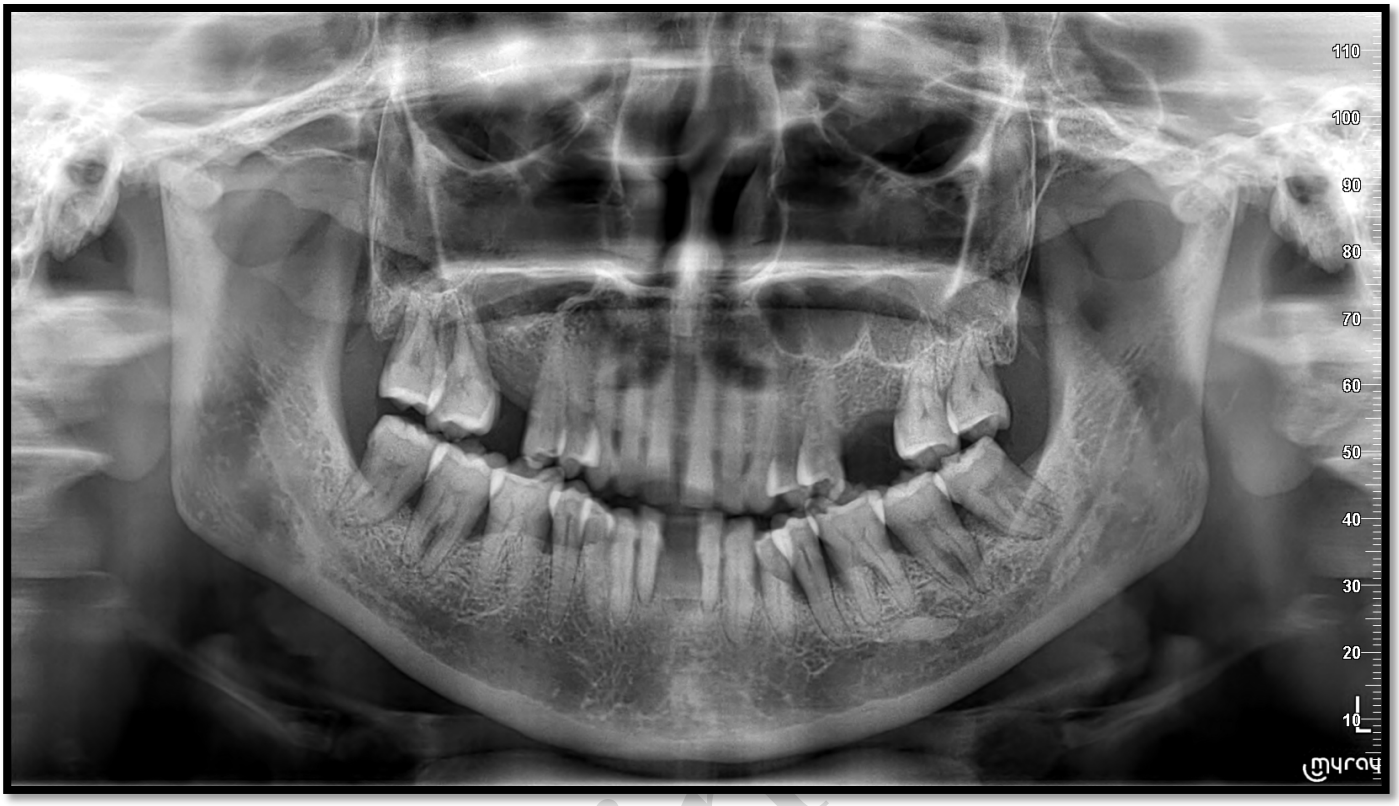

Figure 3: OPG showing generalized periodontitis and missing upper right and left first molars and lower front teeth