

## Diagnostic Sphinx: Sturge Weber Syndrome

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### Abstract

Sturge weber syndrome is a sporadic congenital neuro cutaneous disorders. It is one of the phakomatoses and is often associated with port-wine stains of the face, glaucoma, seizures, mental retardation, and ipsilateral leptomeningeal angioma (cerebral malformations and tumours). Regime dental procedures are associated with increased vulnerability of hemorrhage, making treatment in such patients an imperative task for oral health care practitioners. Unforeseen complications can be prevented by having radical knowledge of this rare syndrome. We hereby report a case of 35 year old female presented with port wine stain and diagnosed as type 2 Sturge-Weber syndrome after investigation.

**Keywords:** Sturge Weber syndrome, Congenital, Encephalotrigeminal angiomatosis, Vascular lesions, Oral manifestations

### Introduction

Sturge weber syndrome (SWS), also known as, meningofacial angiomatosis,<sup>(1)</sup> encephalotrigeminal angiomatosis<sup>(2)</sup> and encephalotrigeminal angiomatosis<sup>(3)</sup> is congenital and non-familial in its origin.<sup>(4)</sup> It belongs to phakomatoses group of disorders (mother-spot diseases).<sup>(4)</sup> It is one of the Systemic syndromes of Phakomatosis pigmentovascularis (PPV)<sup>(5)</sup> and is also categorized into neurocutaneous disorders.<sup>(6)</sup> SWS was first described by Schirmer in 1860 and later more specifically by Sturge in 1879, associated dermatological and ophthalmic changes of the disease to neurologic manifestations. Weber in 1929 complemented it with the documentation of radiologic alterations seen in these patients.<sup>(7)</sup> The increase in the formulation of mutant Gαq protein is due to a somatic activating mutation in guanine nucleotide-binding protein alpha-q (GNAQ). This process is found to be affiliated with SWS.<sup>(8)</sup> It is typically a static lesion but few cases with progressing lesions have been reported.<sup>(9)</sup> Estimated frequency is of 1 per 50,000 live births.<sup>(10)</sup> SWS is characterized by the presence of leptomeningeal angiomas, PWS (sharply demarcated vascular lesions that occur unilaterally along the dermatomes supplied by first two divisions of trigeminal nerve), along with ocular disorders, CNS involvement and oral involvement.<sup>(11)</sup> Manifestation of the syndrome is due to enduring dysfunction of embryonal vascular system<sup>(12)</sup> which develops during the 6th week of embryonic life.<sup>(13)</sup>

Presence of only two of the positive findings i.e. presence of facial angiomas, ocular involvement such as glaucoma, which may or may not be present, suggests diagnosis of Type 2 SWS. Complete absence of CNS involvement is a peculiar finding of type 2 SWS.<sup>(14)</sup> Manifestations such as ipsilateral angiomatosis of lips (causing macrochelia) can be seen. Intraorally, gingival lesion which can be present as vascular hyperplasia or massive hemangiomas proliferation.

Also, pyogenic granulomas may be seen. Unilateral angiomatosis of buccal mucosa, palate, and floor of the mouth is seen on the affected side. Ipsilateral Hypertrophy of alveolus, premature eruption, or delayed eruption and malocclusion are the other abnormalities reported till date.<sup>(15)</sup> Owing to its occasional incidence and a wide spectrum of exemplifications in its manifestations (skin +brain +eyes +oral cavity) we report a case of a female, showing idiosyncratic illustrations of this syndrome.

### Case Report

A 35 year old female reported with a chief complaint of pain and swelling on right mid-face region and presented with pigmentation in the same region, which was static in size since birth. However, she alleged an increase in intensity of color with age from light pink to deep purplish red. Past medical history was negative with respect to any systemic problem, seizure episodes, any drug intake, trauma or abnormality during pregnancy and delivery. No relevant family history was reported. On extraoral examination a maculo-papular area of deep purplish-red pigmentation was noticed over right middle third of face which was unilateral and not crossing the midline. The rash was extending over the supraorbital region, infra-orbital region including bridge of the nose, malar region of cheek and the supralabial region (Fig. 1) along with a gross enlargement of right half of the upper lip suggestive of macrochelia (Fig. 2). Patient didn't report any bleeding, burning sensation, numbness or paresthesia in the pigmented or surrounding areas. Examination of the right eye revealed a visual acuity of no light perception, conjunctival chemosis, episcleral haemangioma with raised intraocular pressure suggestive of glaucoma.(Fig. 3)



**Fig. 1:** Extra oral Photograph showing deep purplish red macular rash on right side of the face.(PWS)



**Fig. 2:** Extraoral photograph showing macrocheilia of right upper lip



**Fig. 3:** Extraoral photograph showing purplish red pigmentation of the right eye

On palpation, absence of bruit or pulsation was noticed. An intraoral examination revealed erythematous, swollen and edematous gingiva involving the right maxillary arch, which was tender on palpation and soft in consistency and restricted within the midline.(Fig. 4) Similar macular purplish red patch was seen involving the right side of the buccal mucosa (Fig. 5) and on hard palate extending to the midline (Fig. 6). Patient's oral hygiene was poor and showed

chronic generalized periodontitis, spacing with maxillary anteriors and prognathic maxilla.(Fig. 4).



**Fig. 4:** Intraoral photograph showing edematous gingiva with purplish red pigmentation on right side



**Fig. 5:** Intraoral photograph showing purplish red macular rash over right buccal mucosa

A Lateral skull and PA radiograph was advised to find out presence of any tram track gyral calcifications but did not reveal any such changes.(Fig. 7 a & b) The haemogram of the patient was well within normal limits. Based upon history and clinical appearance a final diagnosis of type 2 SWS was given. Patient along with her family was educated and made aware about the condition and the possible complications that could arise during the necessary dental procedures. She was prescribed with antibiotics and analgesics course of 5days and advised for oral prophylaxis and extraction with all grossly carious teeth(Fig. 8) followed by replacement with fixed prosthesis.



**Fig. 6:** Intraoral photograph showing unilateral involvement of hard palate with purplish red macular pigmentation



**Fig. 7a, b: Posteroanterior view and lateral skull view radiographs with no radiographic abnormality**



**Fig. 8: Orthopantomograph radiograph showing grossly carious teeth**

## Discussion

According to National Institute of Neurological Disorders and Stroke, SWS is a neurological disorder. It is characterized by a congenital facial birthmark, known as a capillary malformation and other neurological abnormalities. The other symptoms include eye and internal organ irregularities which vary in degree.<sup>(15)</sup> It has no racial or sex predilection.<sup>(16)</sup>

The most apparent sign of SWS is a birthmark or PWS on the face. The PWS is due to an overabundance of capillaries just beneath the surface of the involved areas. When the port wine stain covers the eye and forehead region of the face, SWS should be considered. In rare instances, SWS is present even in the absence of the PWS.<sup>(17)</sup>

SWS can be cited as comprehensive only when both CNS and facial angiomas are present and is considered to be fragmentary when only one area is affected without the other. It can be classified as complete and incomplete depending on presence of facial and leptomeningeal angioma.<sup>(18)</sup> Roach developed a scale for classification which is as follows:<sup>(19)</sup>

Type I - Both facial and leptomeningeal angiomas; may have glaucoma

Type II - Facial angioma alone (no CNS involvement); may have glaucoma

Type III - Isolated leptomeningeal angioma; usually no glaucoma.

The oral manifestations include ipsilateral port-wine stains of oral mucosa along with the hypervascular changes. Angiomatous lesions of gingiva which can vary from slight vascular hyperplasia to hemangiomatous proliferation. It is characterized by increase in the vascular component and gingival hemorrhage at minimal traumatism.<sup>(6,19,20)</sup> Gingival hyperplasia can also be attributed to anticonvulsant medication and secondary to poor oral hygiene in mentally retarded patients. Macroglossia and maxillary bone hypertrophy have also been reported in a few cases.<sup>(21)</sup>

The ocular complications manifests as glaucoma, vascular malformations of conjunctiva, episclera, choroid and retina. This may produce a localized or diffuse pinkish discolouration of the bulbar conjunctiva especially in the limbus zone.<sup>(7)</sup>

Tram track calcification caused by calcification in apposing gyri, ipsilateral calvarial thickening and enlargement of the paranasal sinuses and mastoid may be visible in skull films. Higher imaging modalities like CT for calcification and MRI for brain assessment can also be used. MRI is the current gold standard for diagnosis of this disease especially in infants.<sup>(23)</sup>

Differential diagnosis of SWS includes Rendu Osler Weber syndrome, Von Hippel Lindau Disease and Maffucci Syndrome. Diagnosis is made only on the basis of clinical and imaging features.<sup>(24)</sup>

Hereditary hemorrhagic telangiectasia (HHT) or Rendu Osler Weber Syndrome along with positive family history is characterized by the presence Nosebleeds (epistaxis), mucocutaneous telangiectases and visceral arteriovenous malformation (AVM). AVM's may be pulmonary, cerebral, hepatic, spinal, gastrointestinal and pancreatic. The clinical diagnosis of HHT is considered definite, possible or suspected and unlikely it depends on the number of findings present.<sup>(25)</sup>

Von Hippel-Lindau disease (VHL) is an autosomal dominantly inherited disorder predispose to the development of a variety of tumours (most commonly retinal and central nervous system haemangioblastomas, clear cell renal carcinoma and pheochromocytomas). Visceral cysts (renal, pancreatic and epididymal) are common but rarely compromise organ function. Less frequent tumours include adrenal and extra-adrenal pheochromocytomas, non-functioning pancreatic endocrine cancers, endolymphatic sac tumours and occasionally, head and neck paragangliomas.<sup>(26)</sup>

Maffucci syndrome is characterized by benign enlargements of cartilage (enchondromas); bone deformities; and dark, irregularly shaped hemangiomas. There is a tendency for malignant transformation of enchondromas into chondrosarcomas or of hemangiomas into vascular sarcomas. Patients with Maffucci syndrome also are susceptible to the

development of other malignant lesions such as glioma.<sup>(27)</sup>

Table 1: Demonstrates classical clinical manifestations of SWS and of those present in our case. Our patient exhibited characteristic clinical manifestations of Type 2 SWS including PWS and oral manifestations.

**Table 1: Positive manifestations in the present case of SWS**

Sr. No	Manifestations seen in SWS	Present case findings
1	Epilepsy	-
2	Port Wine Stain	+
3	Abnormal Radiographic Findings	-
4	Mental Retardation	-
5	Oral Manifestations	+
6	Hemiparesis	-
7	Ocular Manifestations	+

Treatment and prognosis of SWS depends on the extent of involvement. PWS can cause severe psychological trauma to the patients and hamper their personality development. PWS can be improved by dermabrasion, tattooing and flash lamp pulse dyed lasers. Various treatment modalities like sclerotherapy, cryotherapy, laser and surgical excision have been tried with varying degrees of success to surmount intraoral lesions.<sup>(28)</sup>

Dental management should be mostly stressed on preventive measures. Patients should be educated, motivated and complied to follow a strict oral hygiene regimen to prevent dental caries and secondary gingival inflammatory enlargement. These patients can undergo endodontic procedure but over instrumentation should be avoided. Gingival hyperplasia has been reported to be successfully managed with CO<sub>2</sub> laser surgery with minimal hemorrhage.<sup>(29)</sup>

It is a challenging task to carry out dental procedures in a SWS patient due to risk of severe intra- and postoperative haemorrhage. Special precautions to keep in mind in order to prevent and treat complications may include hospitalization, application of local anaesthetics with vasoconstrictors, dressings, splints.<sup>(28)</sup>

## Conclusion

Stupendous scope of clinical manifestations of SWS makes diagnosis a critical task. Patients affected with SWS may or may not exhibit intraoral manifestations. It is crucial for oral health care practitioners' to have keen and deep rooted knowledge of this rare congenital disorder and exhibit surplus surveillance during routine dental procedures in order to prevent from life's threatening complication.

## Key Message / Clinical Significance

Although Sturge weber syndrome is a very rare entity, yet it has serious complications. Morbidity can range from minor bleeding to life threatening haemorrhage, hypoxia, infarction, focal deficits, blindness or vascular steal phenomenon. So it is very much necessary to take precautions and to be aware of different types of this syndrome with their manifestations, prior to perform any dental procedure in such patients.

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