Diagnostic Sphinx: Sturge Weber Syndrome

Ashwini Nerkar¹*, Rajeev Gadgil2, Ajay Bhoosreddy3, Karan Shah4, Gaytri Mehroka

¹PG Student, ²Professor, ³Professor & HOD, ⁴Lecturer, MGV’s LBH Dental College & Hospital, Nashik, Maharashtra

*Corresponding Author:
Email: dr.ashwini.nerkar@gmail.com

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. This process is also known as Sturge-Weber syndrome. 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. The condition is often characterized by 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, encephalotrigeminal angiomatosis, and encephalotrigeminal angiomymatosis is congenital and non-familial in its origin. It belongs to phakomatoses group of disorders (mother-spot diseases). It is one of the Systemic syndromes of Phakomatosis pigmentovascularis (PPV) and is also categorized into neurocutaneous disorders. Sturge 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. The increase in the formulation of mutant Gq 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. It is typically a static lesion but few cases with progressing lesions have been reported. Estimated frequency is of 1 per 50,000 live births. SWS is characterized by the presence of leptomeningeal angiomomas, 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. Manifestation of the syndrome is due to endurant dysfunction of embryonal vascular system which develops during the 6th week of embryonic life.

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. Manifestations such as ipsilateral angiomatosis of lips (causing macrochelia) can be seen. Intraorally, gingival lesion which can be present as vascular hyperplasia or massive hemangiomatous 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. 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 paraesthesia 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)
chronic generalized periodontitis, spacing with maxillary anteriors and prognathic maxilla.(Fig. 4).

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.

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
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 traumatisms.\(^6,19,20\) 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.\(^21\)

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.\(^7\)

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.\(^23\)

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.\(^24\)

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.\(^25\)

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 haemangiblastomas, clear cell renal carcinoma and phaeochromocytomas). Visceral cysts (renal, pancreatic and epididymal) are common but rarely compromise organ function. Less frequent tumours include adrenal and extra-adrenal phaeochromocytomas, non-functioning pancreatic endocrine cancers, endolymphatic sac tumours and occasionally, head and neck paragangliomas.\(^26\)

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.\(^{(25)}\)

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**

<table>
<thead>
<tr>
<th>Sr. No</th>
<th>Manifestations seen in SWS</th>
<th>Present case findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Epilepsy</td>
<td>-</td>
</tr>
<tr>
<td>2</td>
<td>Port Wine Stain</td>
<td>+</td>
</tr>
<tr>
<td>3</td>
<td>Abnormal Radiographic Findings</td>
<td>-</td>
</tr>
<tr>
<td>4</td>
<td>Mental Retardation</td>
<td>-</td>
</tr>
<tr>
<td>5</td>
<td>Oral Manifestations</td>
<td>+</td>
</tr>
<tr>
<td>6</td>
<td>Hemiparesis</td>
<td>-</td>
</tr>
<tr>
<td>7</td>
<td>Ocular Manifestations</td>
<td>+</td>
</tr>
</tbody>
</table>

Treatmen 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.\(^{(28)}\)

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\(_2\) laser surgery with minimal hemorrhage.\(^{(29)}\)

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.\(^{(28)}\)

**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.

**References**