CASE REPORT

Sturge Weber Syndrome: An Unusual Case with Multisystem Manifestations

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ABSTRACT

BACKGROUND: Sturge-Weber syndrome (SWS) is a rare congenital neurocutaneous disorder. It is characterized by the presence of facial port wine stains, neurological abnormalities like seizures and mental retardation, ocular disorders, oral involvement and leptomeningeal angiomas.

CASE REPORT: A 13-year-old boy presented with the chief complaint of swollen, bleeding gums and deposits on the teeth. Detailed medical and dental history, clinical examination and investigations confirmed the diagnosis of Sturge-Weber syndrome. The treatment comprised of a thorough plaque control regimen to reduce the gingival enlargement, and it included oral hygiene instructions, thorough scaling, root planing at regular intervals and plaque index scoring which motivated the patient at each visit.

CONCLUSION: This case illustrates that early intervention in a patient with Sturge-Weber syndrome is quintessential because of its associated gingival vascular features and their complicating manifestations. Furthermore, the need for periodic oral examinations and maintenance of good oral hygiene to prevent any complications from the oral vascular lesions has been highlighted.

KEYWORDS: Gingival overgrowth, Hemangioma, Plaque control regimen, Port wine stains, Sturge–Weber syndrome

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INTRODUCTION

Sturge-Weber syndrome (SWS), also known as Sturge- Weber-Dimitri syndrome, encephalotrigeminal angiomatosis or meningofacial angiomatosis, is a rare developmental neurocutaneous disorder that results from dysgenesis of the neural crest in embryologic life (1). Sturge-Weber syndrome occurs at a frequency of approximately 1 per 50,000 people (2). The criteria for the diagnosis of SWS include port wine stains (PWS) of the face (76%), and any combination of seizures (80%), hemiparesis 37%), mental retardation (54%), ocular involvement (37%) or abnormal neuroradiologic findings (63%) (3,4).

The port wine stain is a congenital malformation of the dermis that involves venules, capillaries and possibly perivenular nerves (5). Although port wine stain can occur anywhere in the face, the most common locations in SWS are the V1 and/or V2 areas of the distribution of the trigeminal nerve (6). WS may extend to the neck, the limbs and other parts of the body and can be associated with a hypertrophy of the underlying soft tissues and bones (5,6).

The angioma of the leptomeninges in SWS patients occurs as a unilateral lesion, overlying the posterior temporal, parietal and occipital areas of the brain. The presence of angioma causes leptomeningeal vascular malformation resulting in precipitation of calcium deposits in the cerebral cortex underlying the angioma that is visible as gyriform calcifications in skull radiographs. Seizures, mental retardation, hemiplegia or hemiparesis develops secondary to this process and their severity depends on the extent of the lesion (3). Ocular involvement is pathognomonic

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and can result in glaucoma, choroidal haemangioma, bupthalmos or hemianopsis (3).

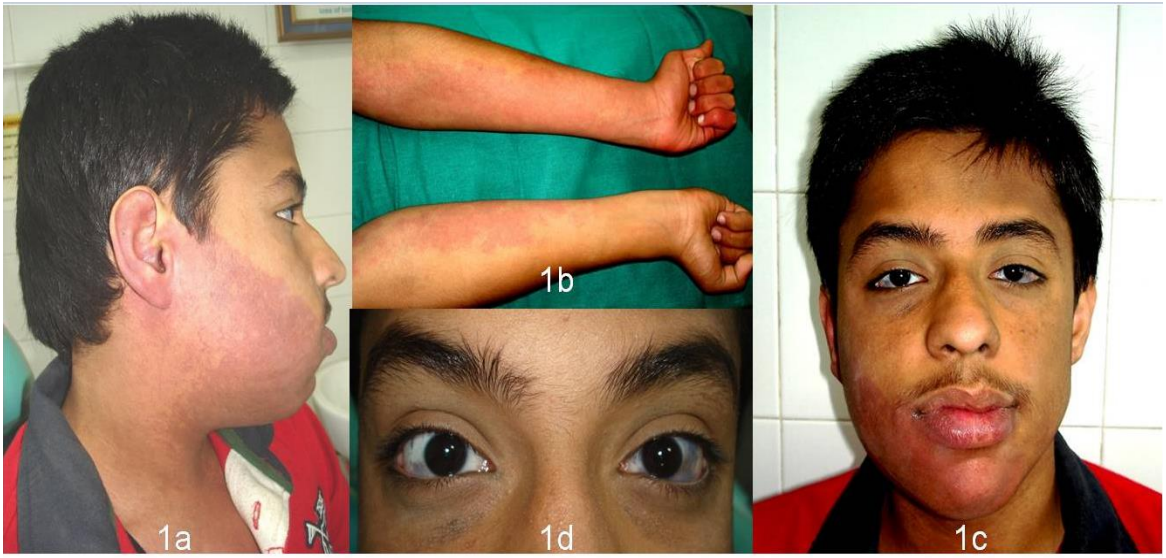
Intra orally, angiomatosis may involve buccal mucosa, palate, gingiva, floor of the mouth and lips, causing macrochelia. Gingival enlargement can vary from light vascular hyperplasia to monstrous overgrowth. These gingival lesions make closure almost impossible and are disfiguring. In addition, impairment of nutrition and oral hygiene can be caused by the moderate to severe forms of gingival overgrowth, leading to increased susceptibility to oral infections and deterioration of the systemic health. Other reported intraoral abnormalities associated with SWS are pyogenic granulomas, unilateral hypertrophy of alveolus, ipsilateral premature eruption or delayed eruption and malocclusion (7). Sturge-Weber syndrome is of rare occurrence, and the management becomes complicated due to the risk of hemorrhage. Although the syndrome has been completely described, little has been mentioned in the literature about the oral and dental care. The present case report intends to describe the clinical and radiographic features in a SWS patient, the dental treatment that was provided and the follow up results.

CASE REPORT

A 13-year-old male patient reported for the evaluation and treatment of swollen, bleeding gums and deposits on the teeth.

History: Medical history depicted that the patient had tonic clonic convulsions at five years of age, and that he was treated with phenobarbitone medicine. For the past seven years, he was free of epileptic symptoms and was not on medication either. There was no sign of mental retardation, and the patient was communicative. The family history did not reveal any similar complaint from his immediate or distant relatives. The patient was the elder of the two siblings, born at full term by cesarean delivery. History also revealed that the reddish discoloration was present on the face and other parts of the body since birth and was gradually darkening with age.

Extra oral examination: Extra oral examination showed that the port wine stains were distributed on the right side of the face, ear, the neck and both the arms [Figures 1a, 1b]. Facial asymmetry and macrochelia were noted, with enlargement on the right-side [Figure 1c]. Examination of the eyes showed that the blood vessels in both eyes were dilated [Figure 1d], and the patient was advised ophthalmic consultation.



Figures 1a,1b*: Unilateral distribution of the portwine stains on the face, ear, neck, right and left arms*
Figure 1c*: Photograph revealing facial asymmetry and macrochelia, with enlargement on the right side*
Figure 1d*: Dilated blood vessels of both eyes*

Intra oral examination: Intra oral examination revealed reddish discoloration of buccal mucosa, floor of the mouth, palate and gingiva. Gingival enlargement was prominent on the upper anterior region and posteriorly on the right side**.** The enlarged gingiva showed blanching on pressure suggesting angiomatous enlargement. Midline shift of the dentition on the left side was seen along with presence of deep bite and cross bite in relation to 33 and 44. The oral hygiene of the patient was poor, with extensive amounts of plaque and calculus present which might have aggravated the condition [Figures 2,3].

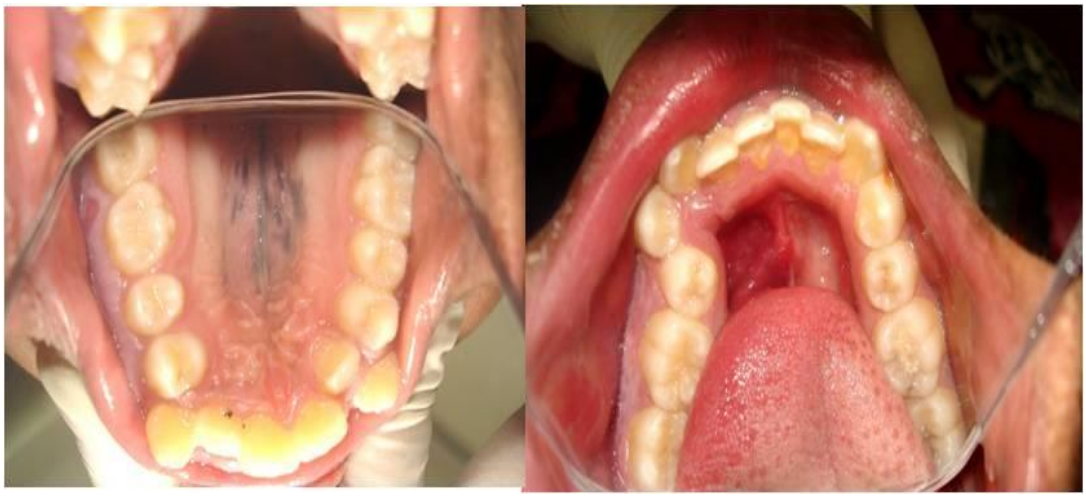


Figure 2: *Photograph showing the reddish purple discoloration on the palate and angiomatous lesion on the floor of the mouth*

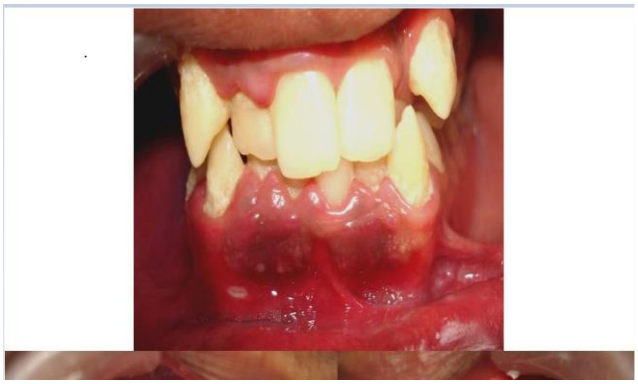


Figure 3*: Gingival enlargement in upper anterior region, posterior right region and normal gingiva on the left posterior region*

Radiographic examination: MRI of the face was performed using spin echo and fast spin echo pulse sequences and serial T1. IR weighted images were also obtained in the axial, coronal and sagittal planes. TI weighted spin echo images were repeated in the sagittal, axial and coronal planes following administration of intravenous contrast (Gd-DPTA). Altered signal area due to haemangioma was noted involving pterygoid muscle and right parapharyngeal space. Subtle hyperintensity on IR images were noted involving ramus of the mandible on right side, and the mild expansion of the right body of the mandible was seen [Figure 4]. Lateral skull showed no evidence of intracranial calcifications [Figure 5].

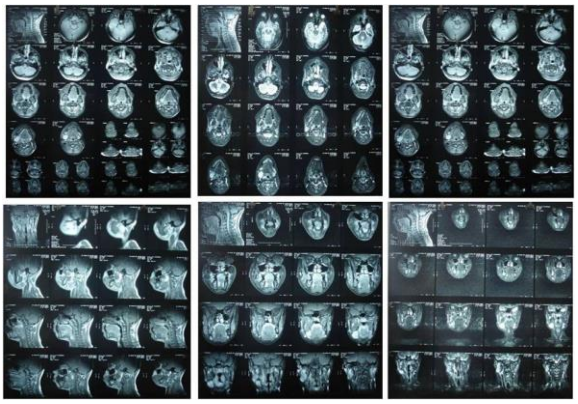


Figure 4*: MRI of the face depict haemangioma involving pterygoid muscle, right parapharyngeal space and mild expansion of right body of mandible*

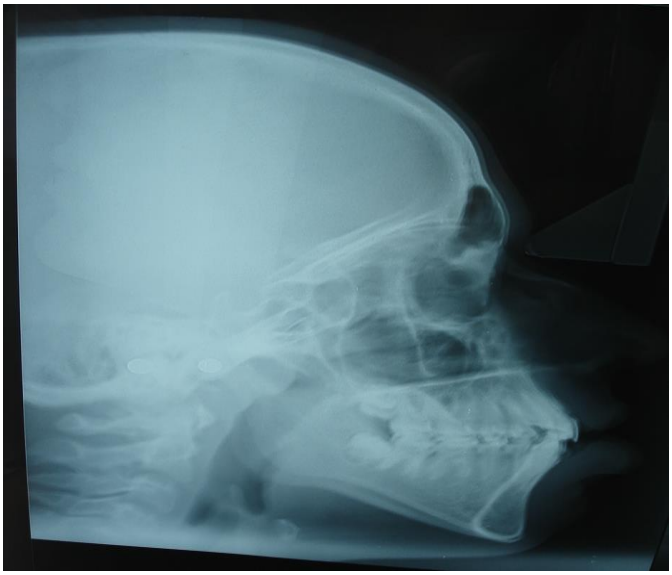


Figure 5*: Lateral skull radiograph showing no intracranial calcifications*

Based on clinical and radiographic features, diagnosis of Sturge-Weber syndrome was made. The treatment procedures were explained to the patient and his parents, and a written informed consent was obtained from them. Physicians' consent was obtained prior to thw treatment.

Treatment: The main therapeutic goal was to improve the periodontal condition of the patient. A thorough plaque control regimen was followed to reduce the gingival enlargement. It included deep scaling, root planing at regular intervals, plaque index scoring and motivating the patient at each visit [Figure 6a].

The patient and his parents were educated regarding good oral hygiene maintenance practices. In addition the patient was advised to use 0.2% chlorhexidine mouthrinse after brushing.

Follow- up: At scheduled follow-up visit after one month, the patient reported that no bleeding occurred during brushing and that the degree of the gingival enlargement had also decreased marginally [Figure 6b]. The patient was followed up every three months, for two years, to evaluate plaque control, look for signs of recurrence of gingival enlargement and continue with the plaque control regimen.

Reevaluation of the patient after three months showed remarkable reduction of the gingival enlargement in the maxillary and mandibular arches. Close follow-up and complete plaque control were done every three months, for a period of two years showed a remarkable improvement in the periodontal condition of the patient [Figure 6c].

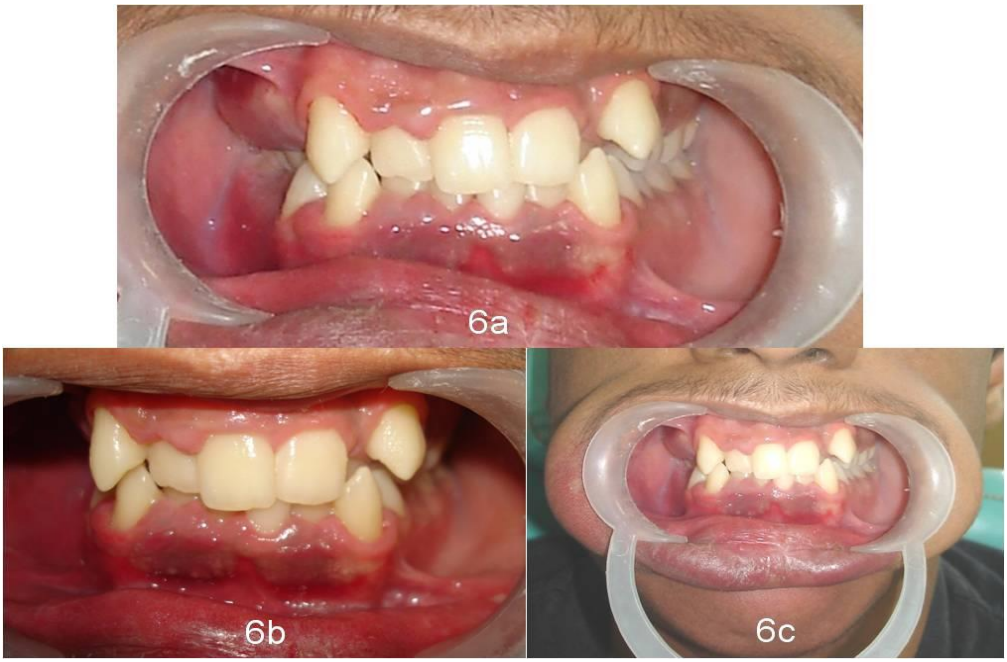


Figure 6a, 6b, 6c*: Postoperative view after plaque control, at follow up visit after 1 month of treatment and after 2 years of treatment*

DISCUSSION

Although the exact etiology of Sturge-Weber syndrome is unknown, it is believed that the diverse clinical features are a result of a common embryologic origin. SWS is caused by an abnormal persistence of an embryonal vascular system, localized around the cephalic portion of neural tube. This vascular plexus normally forms at the 6th week of intrauterine life and regresses at the 9th week. Failure of its regression results in residual vascular tissue, which forms angiomas of leptomeninges, face and eyes (8).

Differential diagnosis of Sturge-Weber syndrome includes Von Hippel Lindau disease (a familial syndrome involving hemangioblastoma in cerebellum, retina and renal cysts), angio-osteodystrophy syndrome (port wine stains in face, varices and hypertrophy of bone), Render Osler-Weber syndrome (abnormal dilatation of terminal vessels of skin, mucosa and viscera) and Maffuci's syndrome (multiple angiomas of skin and chondromas of bone) (3). Port wine stains should be differentiated from pyogenic granuloma, nevus flammeus neonatorum and venous varicosities (9).

Treatment and prognosis of the patient depend on the nature and severity of clinical features. Port wine stains on the face can cause deep psychological trauma to the patient and should be treated by a plastic surgeon by dermabrasion, tattooing, sclerotherapy, photo coagulation by lash lamp pulse tunable dye lasers and a combination of these. Antiepileptic treatment for seizure control and the medical, surgical management of glaucoma form the mainstay of the treatment. Cryosurgery may be used to correct lip and other soft-tissue deformities (10,11).

Intraoral involvement is common, resulting in hypervascular changes to the ipsilateral mucosa (3). The subject in the present case illustrated a massive hemangiomatous proliferation of the gingiva that was soft on palpation and showed blanching effect. The modalities that have been used for the treatment of gingival enlargement include conscientious observation, radiation therapy, steroids, antimetabolites, injecting sclerosing solutions into the gingival tissue and surgical removal of the gingival growth with electrosurgery, cryosurgery, or lasers (12). The dental rehabilitation of patients with Sturge-Weber syndrome is a challenging task and requires an initial conservative management and later a surgical intervention (13). Dental education regarding the oral hygiene practices and preventive strategies should be provided to the patient and parents as was done in this case. This can avoid gingivectomy, which can be quite threatening in these patients (14). The risk of hemorrhage in Sturge-Weber patients should really reinforce the importance of good dental health for affected patients.

Despite the strict oral hygiene measures, if gingivectomy is mandatory, then the patient must be hospitalized since achieving hemostasis can be a major problem. The risk of hemorrhage can be managed by the use of haemostatic agents like topical bovine thrombin, use of postoperative splints, injecting sclerosing solutions, percutaneous transcatheter vascular embolization using gelfoam or polyvinyl alcohol and provision for blood transfusion (15).

Nd-YAG laser used in a contact mode with a fiber-optic hand piece can be an efficient option to perform gingivectomy in patients with Sturge-Weber syndrome. Its advantages include: achievement of immediate hemostasis, minimal damage to the surrounding tissues leading to no postoperative pain, attaining a sterile field and thus, no risk of secondary infection and no requirement for postoperative periodontal dressing. Wilson et al recommended the use of a positive pressure splint to prevent recurrence of hemangiomatous growth following gingivectomy (16).

Orthodontic treatment of Sturge-Weber patients can be affected by the alveolar overgrowth, gingival enlargement and poor oral hygiene (11). Malocclusion, problems in speech and trauma during mastication can be associated with macroglossia, which was also seen in this case. Macroglossia can be managed in these patients with partial glossectomy combined with lasers (17).

In conclusion, in the present case, treatment outcomes were favourable. The ase illustrates the need for an appropriate and timely intervention in a patient with Sturge-Weber syndrome because of its associated gingival vascular features and their complicating manifestations. Periodic oral examinations and maintenance of good oral

hygiene helped to prevent any complications from the oral lesions and to improve the quality of life of the patient with sturge-weber syndrome.

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