CASE REPORT

Sturge–Weber Syndrome: Roots to a Cure a Nightmare in Pediatric Dentistry

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Abstract

Aim and objective: This article aims to report a case of Sturge–Weber syndrome (SWS) in a pediatric patient and its systematic dental management to add to the existing literature.

Background: Sturge–Weber syndrome is one of the vascular disorders, known for its stupendous scope of clinical manifestations and life-threatening complications. The substantial prevalence of oral manifestations in SWS makes it crucial to have a comprehensive knowledge of this rare congenital disorder.

Case description: This case report represents a case of SWS in an 11-year-old boy with oral, ocular, and neurological features who reported due to pain, unilateral gingival enlargement associated with spontaneous bleeding in the mandibular left region. A multidisciplinary team approach having comprehensive knowledge regarding such rare congenital disorder is a must to prevent its life-threatening complications.

Conclusion: Sturge–Weber syndrome often affects the oral cavity through vascular lesions. Hence, deep knowledge is immensely important to provide an appropriate dental treatment without complications.

Clinical significance: Port-wine stains should not be considered as just birthmarks and should be further investigated for its systemic involvement to arrive at a confirmatory diagnosis and treated accordingly with special precautions.

Keywords: Computed tomography head, Dental management, GNAQ gene, Pediatric dentistry, Port-wine stains, Tranexamic acid.

International Journal of Clinical Pediatric Dentistry (2021): 10.5005/jp-journals-10005-1928

Background

Hemorrhagic disorders are relatively rare genetic disorders characterized as the inability to form a blood clot that leads to abnormal spontaneous bleeding or profuse bleeding after the mildest provocatory event. Bleeding disorders are broadly categorized as deficiency of coagulation factors, platelet disorders, and vascular or fibrinolytic defects.[1] Vascular defects are sparser in occurrence, usually associated with bleeding confined to skin or mucosa. Vascular defects are ordinarily marked by the presence of birthmarks.[2] Sturge–Weber syndrome (SWS) is one of the vascular disorders, known for its stupendous scope of clinical manifestations and life-threatening complications makes it a critical and mandatory task to arrive at its accurate diagnosis. It is a congenital disorder, sporadic in origin and it is linked to syndromes of phakomatoses disorders (mother-spot diseases). Nearly a century ago (1860), Schirmer was the first one to describe SWS in alliance to angioma of face and buphthalmos. William Allen Sturge in 1879 highlighted dermatological and ophthalmic manifestations of SWS which were escorted by radiological alterations put forth by Weber.[3,4] Anticipated occurrence of SWS is 1 per 50,000 live births.[5] Sturge–Weber syndrome, also known as encephalofacial angiomatosis and encephalotrigeminal angiomatosis. It is among the neurocutaneous syndromes (NCS) characterized by a pathognomonic triad of facial cutaneous vascular nevus [nevus flammeus or port-wine stain (PWS)], neurological dysfunction, and ocular menifestatations.[6,7]

In 1992, Roach categorized SWS into three variants. Type I: Individual with a PWS on the face, leptomeningeal angioma, may have glaucoma. Type II: Individual with a facial PWS, absence of leptomeningeal angioma, and may have glaucoma. Type III: Individual with leptomeningeal angiomatosis, absence of facial PWS, and, rarely, glaucoma.[8] Intraorally, angiomatosis can outstretch to the buccal mucosa, palate, tongue, the floor of mouth, gingiva, and lips may show purplish-red discoloration. Gingival lesions vary from slight vascular enlargement to massive gingival growths.[9] Oral vascular malformation accounts for approximately 40% of SWS patients.[10] Routine dental and oral surgical procedures seem complicated due to the known risk of intra- and postoperative hemorrhage. The considerable prevalence of oral manifestations of SWS makes it crucial to have comprehensive knowledge about this rare congenital disorder.

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How to cite this article: Mapara PN, Taur SM, Hadakar SG, *et al.* Sturge–Weber Syndrome: Roots to a Cure a Nightmare in Pediatric Dentistry. Int J Clin Pediatr Dent 2021;14(1):145–148.

Source of support: Nil

Conflict of interest: None

Case Description

An 11-year-old boy reported the chief complaint of pain in the lower left back tooth region of the jaw and spontaneous bleeding through the oral cavity past 2 months. The patient gave a history

of few epileptic episodes that occurred at an early age only. The natal history was non-contributory. General examination showed appropriate motor and speech function and absence of cardiovascular or respiratory diseases. Extraorally purplish-red pigmentation patches, i.e., PWS were present and confined to the middle and lower left side of the face without crossing the midline (Fig. 1).

Intraoral examination revealed an extension of erythematous reddish-pink patches of residual vascular plexus on the left side of the oral cavity involving the lower lip, left labial, and buccal vestibule and floor of mouth (Fig. 2). The mandibular left first primary molar was the tooth of chief complaint which was grossly carious and was associated with gingival hyperplastic growth. The gingival hyperplastic tissue tended to bleed profusely even on mild probing. Occlusal caries was present with 54, 55, 65, 75, 84, and 85. An intraoral periapical radiograph (IOPAR) with 74 and orthopantomogram (OPG) was taken to confirm any root resorptions, eruption status of developing 1st premolar and osteohypertrophy (Figs 3 and 4). He had poor oral hygiene, cariogenic diet habits that aggravated the spontaneous bleeding tendency on the affected site due to vascular dysfunction. The observed findings were indicative toward a provisional diagnosis of the case as SWS. The patient has advised computed tomography (CT) scan, radiographic examinations of the head region, complete hematological examinations, and ophthalmic examination. Dermatology, pediatric, and neurology consultations were given to have an accurate diagnosis and to obtain medical fitness of the child to undergo required dental treatment.

Intraoral periapical radiograph with 74 and OPG revealed no evidence of any osteohypertrophy thus confirming only soft tissue gingival hyperplastic growth associated with grossly carious 74 with spontaneous bleeding tendency which was indicated for extraction (Fig. 5). Computed tomography head examination showed a positive finding by the presence of calcifications in the choroid plexus of the brain (Fig. 6). Ocular examination revealed no significant findings but the patient has been advised to follow regular ophthalmic check-ups. The hemogram of the patient was normal. The patient underwent a plaque control regimen that included oral prophylaxis, use of chlorhexidine mouth rinses, and patient–parent education to motivate them to follow a strict oral hygiene routine. Age-appropriate brushing technique was demonstrated and diet counseling was done. Glass ionomer cement restorations were done with 55, 65, 75, 84, and 85. Pit and fissure sealant application was done with 16, 26, 36, and 46. Topical fluoride application was done. Extraction was indicated with 54 and 74. On completion of restorative and preventive

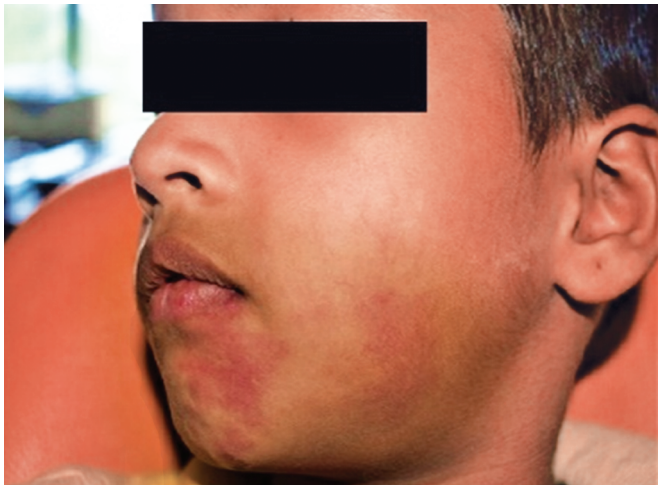

Fig. 1: Port-wine stains present on the left side of the face

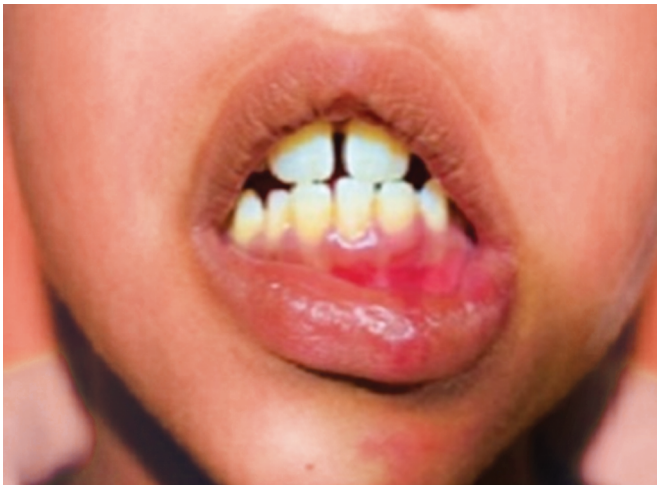

Fig. 2: Intraoral extension of port-wine stains

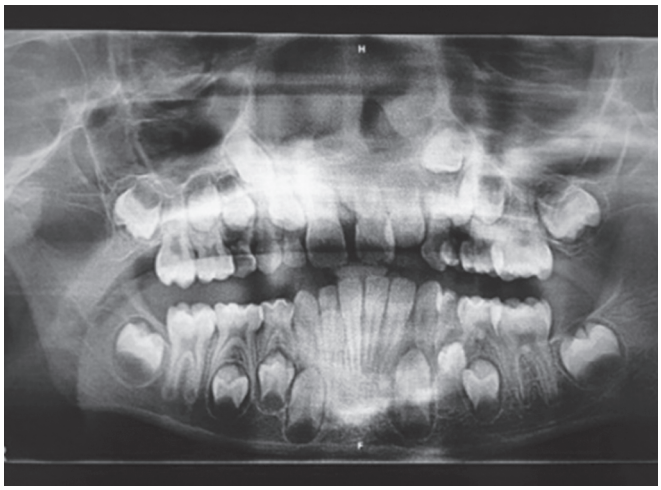

Fig. 3: Orthopantomograph shows grossly carious mandibular left 1st primary molar (74)

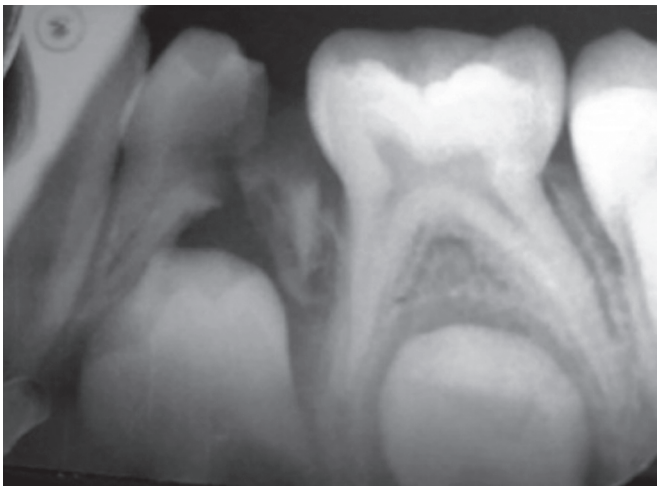

Fig. 4: Intraoral periapical radiograph shows mandibular left 1st primary molar (74)

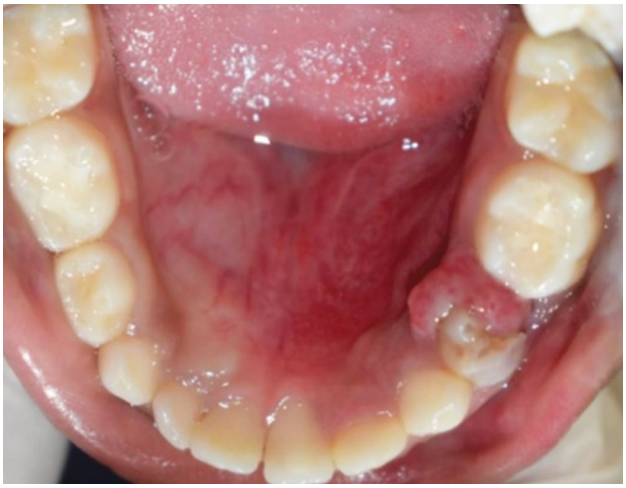

Fig. 5: Gingival growth associated with 74 with a tendency to bleed profusely due to excessive residual vascular plexus in the vicinity of 74 indicated for extraction. The picture also shows the completion of other preventive and restorative dental treatments

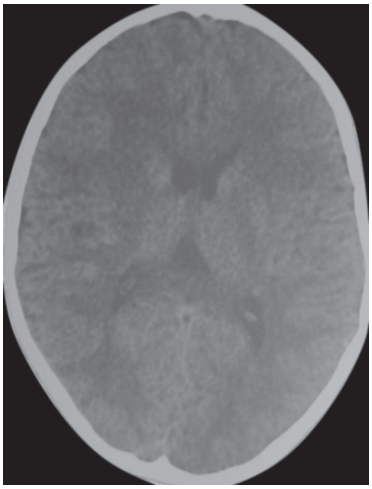

Fig. 6: CT head shows calcifications in choroid plexus of the brain

Table 1: Positive manifestations in the present case of Sturge–Weber syndrome

S. no.	*Manifestations seen in SWS*	*Present case findings*
1	Epilepsy	+
2	Port-wine stain	+
3	Abnormal head radiographic findings	+
4	Mental retardation	–
5	Oral manifestations	+
6	Ocular manifestations	–

dental treatments, the extraction of 74 was carried out under 2% lignocaine local anesthesia along with excision of associated gingival overgrowth with 74. Careful curettage was performed for granulation tissue removal followed by irrigation of the extraction socket. Complete hemostasis was achieved with a pressure pack of gauze pieces soaked in tranexamic acid. After treatment, the patient was under observation for 3 hours before discharge. Appropriate postoperative instructions were given.

On the follow-up visit again emphasis was on patient–parent education and motivation to follow strict oral hygiene and a healthy balanced diet regimen. Preventive dental treatments were performed. Follow-up intraoral periapical radiograph was taken to evaluate the development status of the left mandibular 1st premolar tooth which showed adequate development. The patient is under regular follow-up examinations in neurology and ophthalmic specialties with a good prognosis rate due to his early diagnosis done during his dental visit and timely referral of the patient.

Discussion

Sturge–Weber syndrome is a developmental anomaly of embryonic origin occurring as a result of residual embryonal blood vessels. The trademark of the sixth to the ninth week of embryonic life is the development of crucial tissues of the cephalic region of the embryo-like central and peripheral nervous system, sclera of eyes, and facial skin marked by the migration of neural crest cells. As a general rule, the vascular plexus develops surrounding the cephalic portion of the neural tube, under ectoderm decided by the fate to become facial skin in the sixth week and degenerates near the ninth week of gestation. Omission of destined regression leads to residual vascular plexus resulting in the excessive capillary network forming the angiomata of the leptomeninges, face, and ipsilateral eye.[11] The pathognomonic triad of SWS is facial cutaneous venous dilation, also known as a PWS, neurological dysfunction due to leptomeningeal angiomatosis and ocular involvement. A PWS is visible at birth along with the distribution of one of the first developing nerve, i.e., trigeminal nerve. The endurant dysfunction of an embryonal vascular system results in angiomas due to residual vascular plexus in the brain and eye. A "vascular steal phenomenon" may develop around the angioma in the leptomeninges of the brain leading to pooling of blood and obstruction to venous return creating difficulty in the entry of fresh blood to affected regions of the brain which suffers ischemic insult, undergoes atrophy, and shows calcifications. Neurological dysfunction such as recurrent seizures, status epilepticus, intractable seizures along with recurrent vascular events can propagate this steal further, with an elevation in cortical ischemia, resulting in progressive calcification, gliosis, and atrophy leading to seizures and neurological deterioration. The cardinal ophthalmic manifestations (i.e., buphthalmos, glaucoma) occur secondary to elevated intraocular pressure (IOP) along with mechanical obstruction of the angle of the eye, elevated episcleral venous pressure, or marked secretion of aqueous fluid.[12,13]

Advanced diagnostic techniques like CT for calcification and magnetic resonance imaging (MRI) for brain assessment are considered to be gold standard methods.[14] Differential diagnosis of SWS includes nevus flammeus, Rendu–Osler–Weber syndrome, Von Hippel–Lindau disease, and Maffucci syndrome. Diagnosis is accurately done only based on clinical and imaging features.[15]

Table 1 illustrates cardinal manifestations of SWS and manifestations recorded in the present case which indicates type I SWS.

Sturge–Weber syndrome is associated with the risk of severe intra- and postoperative hemorrhage. The pivotal complication after tooth extraction in such patients is whether hemostasis can be achieved when profuse bleeding occurs in the adjacent angiomatotic soft tissue.[16] More often than not, patients with SWS have compromised oral hygiene owing to fear of profuse bleeding from the hyperplasic gingiva affected by angiomatosis while brushing the teeth.[17] The poor oral hygiene favors the exacerbation of bleeding tendency of the affected tissue. Overgrowth of the

gingiva amplifies the gingival inflammation and creates difficulty in maintaining proper oral hygiene.

Before planning a tooth extraction in SWS cases, an understanding of whether the gingiva surrounding the target tooth includes angiomatous tissue is crucial. Some case reports have advocated standard extraction of the tooth surrounded by angiomatous tissue that was performed successfully under local anesthesia without serious hemorrhagic complications. Hemostasis in such cases was achieved only with pressure packing.[18]

The well-being of the patient must be prioritized over the necessity to extract teeth surrounded by angiomatotic tissue as SWS shows serious oral complications with a risk of life-threatening intra- and postoperative hemorrhage. Consequently, it is cardinal to take precautions and to be aware of various consequences in SWS with their manifestations, before performing any dental procedure. The various methods can be used to manage the risk of hemorrhage like patient's blood typed and cross-matched, provision for blood transfusion, use of hemostatic agents, injecting sclerosing solutions, percutaneous transcatheter vascular embolization using gel-foam or polyvinyl alcohol.[19]

Tranexamic acid is known for its safe and effective hemostatic action in managing postoperative hemorrhage in patients undergoing anticoagulant therapy. It is a synthetic variant of lysine that employs its antifibrinolytic mechanism of action via reversible blockage of lysine-binding site on plasminogen molecules leading to its hemostatic mechanism.

Management of SWS is based on the extent of involvement of tissues. The severe psychological trauma of PWS can affect a patient's personality development. Port-wine stain can be treated with dermabrasion, tattooing, and flash lamp pulse-dyed lasers. Different treatment techniques like sclerotherapy, cryotherapy, laser, and surgical excision have been carried out with varying degrees of success to vanquish intraoral lesions.[19]

Dental management is ought to be emphasized on preventive measures. Patients must be well educated and motivated to follow a strict oral hygiene routine to prevent dental caries and secondary gingival inflammatory enlargement. It is a difficult task to perform dental procedures in an SWS patient owing to the risk of severe intra- and postoperative bleeding. An important precaution to keep in mind is that a suitable armamentarium must be at stat available in case any complication emerges. A multidisciplinary approach is the core of the proper treatment of SWS patients.

Conclusion

Sturge–Weber syndrome often manifests as perioral and intraoral vascular lesions. Hence, comprehensive knowledge is crucial to provide an appropriate dental treatment without complications. Information regarding SWS's signs and symptoms as well as the knowledge of the best treatment to be performed will provide an overall health benefit to patients.

Clinical Significance

Port-wine stains should not be considered as just birthmarks and should be further investigated for its systemic involvement to arrive at a confirmatory diagnosis and treated accordingly with special precautions.

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