Open Access Library Journal
2026, Volume 13, e14802
ISSN Online: 2333-9721
ISSN Print: 2333-9705

Sturge Weber Syndrome Revealed by Severe Generalized Gingival Enlargement in a 12-Year-Old Patient: A Case Report

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How to cite this paper: El Khalifa, K., El Bouhairi, M., Adnane, S. and Kissi, L. (2026) Sturge Weber Syndrome Revealed by Severe Generalized Gingival Enlargement in a 12-Year-Old Patient: A Case Report. *Open Access Library Journal*, **13**: e14802.
https://doi.org/10.4236/oalib.1114802

Received: December 23, 2025
Accepted: March 20, 2026
Published: March 23, 2026

Abstract

Background: Sturge Weber syndrome (SWS) is a rare, sporadic neurocutaneous disorder caused by somatic mutations in the GNAQ gene, characterized by vascular malformations involving the skin, leptomeninges, and eyes. Although facial port-wine stains, seizures, and ocular abnormalities are the hallmark features, oral manifestations are less frequently reported and may represent an early clinical sign of the disease. Case Presentation: We report the case of a 12-year-old girl who presented with progressive generalized gingival enlargement associated with spontaneous bleeding and functional impairment. Clinical examination revealed bilateral facial port-wine stains extending to the trunk and limbs, facial asymmetry, and intraoral diffuse hypervascular gingival proliferations affecting all quadrants. Radiographic evaluation showed horizontal bone loss of the mandibular incisors. Multidisciplinary assessment, including dermatology, neurology, and ophthalmology, supported the diagnosis of SWS, which was confirmed by brain magnetic resonance imaging demonstrating leptomeningeal angiomatosis, consistent with SWS type I. Initial management included periodontal therapy, surgical excision of gingival lesions, and tooth extractions under strict hemorrhagic precautions. Histopathological examination revealed a vascular epulis. Despite initial improvement, the patient presented two years later with a severe recurrence of gingival overgrowth, likely related to antiepileptic therapy and inadequate oral hygiene, necessitating referral for comprehensive surgical management. Conclusion: This case emphasizes that vascular gingival overgrowth may represent an early and revealing manifestation of SWS. Dentists play a key role in the early detection of systemic vascular disorders through careful oral examination, enabling timely multidisciplinary diagnosis and management while minimizing hemorrhagic risks.

Subject Areas

Dentistry

Keywords

Sturge Weber Syndrome, Gingival Overgrowth, Oral Manifestations, Vascular Epulis, Port-Wine Stain, Leptomeningeal Angiomatosis, Pediatric Dentistry, Oral Surgery, Multidisciplinary Care

1. Introduction

Sturge Weber syndrome (SWS) is a rare, sporadic neurocutaneous disorder caused by somatic mutations in the GNAQ gene, characterized by vascular malformations affecting the skin, leptomeninges, and eyes [1] [2]. While facial port-wine stains, seizures, and ocular complications are the hallmark features, oral manifestations such as gingival vascular overgrowth are less commonly reported and may serve as an early clinical clue [2]. These lesions can be exacerbated by antiepileptic therapy and poor oral hygiene, increasing the risk of bleeding and functional impairment [3]. Early recognition of oral signs by dental practitioners is therefore critical for prompt multidisciplinary evaluation and management. This report describes a 12-year-old patient in whom severe generalized gingival enlargement led to the diagnosis of SWS, underscoring the role of dentists in detecting systemic vascular syndromes through local manifestations.

2. Case Report

A 12-year-old girl, accompanied by her father and aunt, presented to the Oral and Maxillofacial Surgery Department on November 10, 2022, with a chief complaint of progressive generalized gingival enlargement interfering with mastication and causing spontaneous bleeding. Her past medical history included recurrent untreated epileptic seizures during childhood and mild cognitive delay, with no prior systemic treatment. The patient also reported difficulty maintaining oral hygiene and occasional pain. The history of the present illness indicated long-standing gingival enlargements that had progressively increased over time, with a marked exacerbation two months prior to consultation, leading to significant functional discomfort and frequent spontaneous bleeding.

Extra-oral clinical examination revealed bilateral facial port-wine stains (plan angioma) extending to the trunk and limbs, facial asymmetry, left-sided strabismus, and no cervical lymphadenopathy (**Figure 1**).

Intra-orally, multiple erythematous, soft, compressible, and bleeding gingival proliferations were observed across all quadrants of the maxilla and mandible, ranging from sessile to pedunculated with a red to violaceous coloration suggestive of hypervascularization. Additional findings included mobility of the mandibular incisors, and markedly inflamed surrounding mucosa (**Figure 2**).

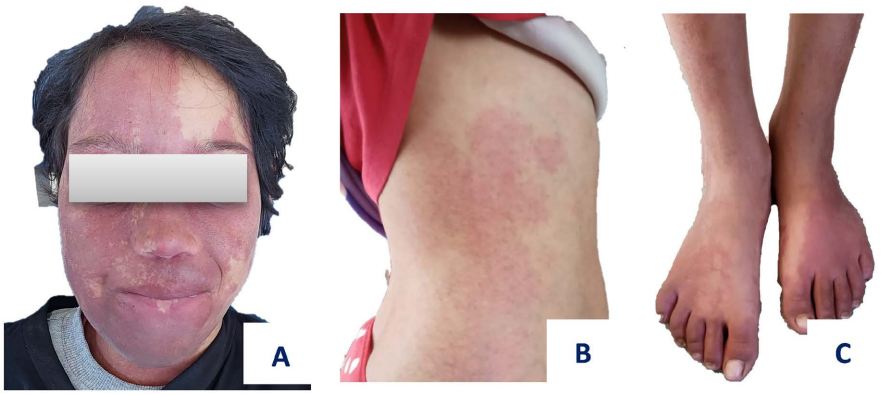

Figure 1. The findings of extra-oral examination. (A) bilateral facial port-wine stains, facial asymmetry, left-sided strabismus hidden for the patient privacy; (B) Showing the trunk port-wine stains; (C) Showing the limbs port-wine stains.

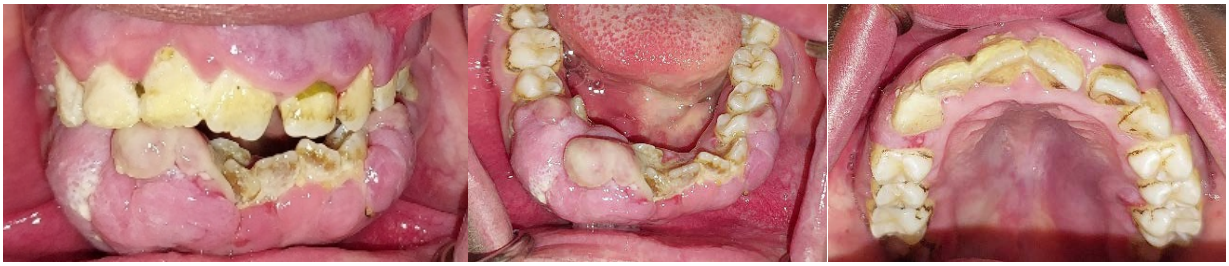

Figure 2. Findings of intra-oral examination.

Radiographic assessment demonstrated horizontal bone loss around the mandibular incisors on panoramic radiograph (**Figure 3**). Taking in consideration all our findings, we suspected that the patient might have Sturge Weber Syndrome (SWS). Specialist consultations in dermatology, neurology, and ophthalmology departments supported our suspicion, considering the history of epilepsy with neurocognitive delay, facial and truncal angiomas, ophthalmologic involvement including strabismus and the right eye glaucoma. The Magnetic Resonance Imaging (MRI) of the brain findings were compatible with SWS, as they show leptomeningeal angiomatosis leading to initiation of antiepileptic therapy with valproic acid and confirmation of SWS Type I (**Figure 4**).

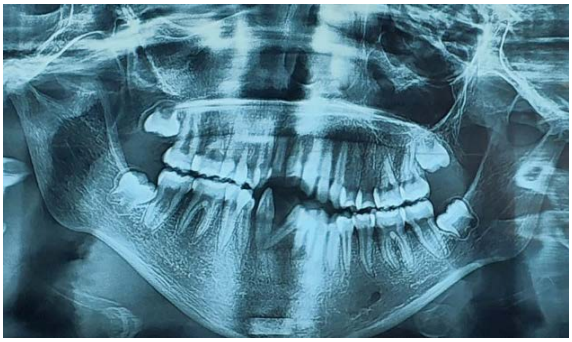

Figure 3. Panoramic radiograph showing horizontal bone loss around the mandibular incisors.

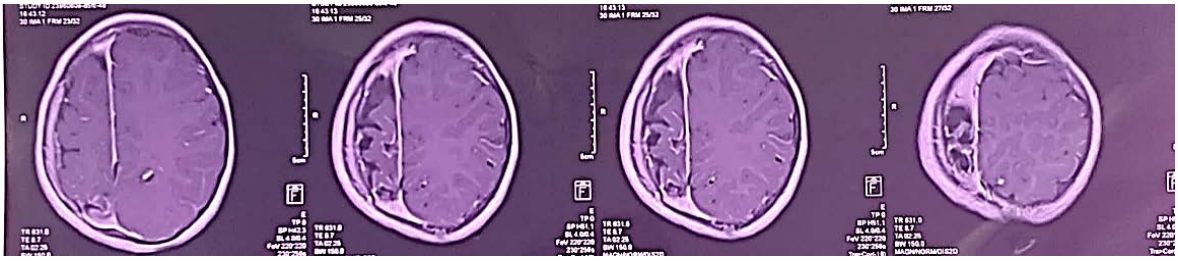

Figure 4. Magnetic Resonance Imaging of the patient's brain showing leptomeningeal angiomatosis.

The treatment plan included oral hygiene instructions and motivation, full-mouth scaling, gingival biopsy, extraction of teeth 42 and 43 due to poor prognosis, and surgical gingival excision. Precautions regarding the hemorrhagic risk were taken: the diode laser was kept within reach, although it was ultimately not required, as hemostasis was achieved using simple compression and hemostatic sponges with sutures (**Figure 5**).

The complete treatment plan was thoroughly explained to the patient and her father. Prior to initiating the procedures, written informed consent was obtained from the parents, and medical clearance was provided by the patient's neurologist regarding the hemorrhagic risk.

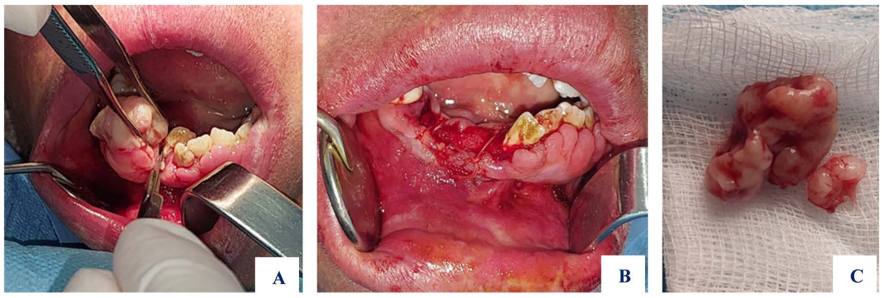

Figure 5. Surgical management of gingival lesions on February 2023. (A) Surgical excision; (B) Immediate postoperative view showing the placement of hemostatic sponges and interrupted sutures used to achieve and stabilize hemostasis; (C) Excised gingival tissue specimen.

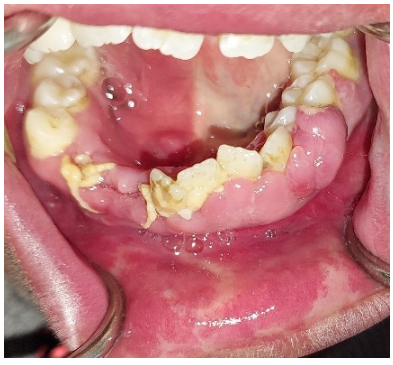

Figure 6. Follow up one month later.

Histological examination showed variably hyperplastic or ulcerated stratified

squamous epithelium overlying edematous connective tissue rich in congested ectatic vessels and mild polymorphous inflammatory infiltrate. No malignancy was observed. These Findings were consistent with a vascular epulis. Following initial periodontal therapy, significant reduction in inflammation was noted. An improvement was observed during follow-up one month after surgery (**Figure 6**).

Unfortunately, the patient was subsequently lost to follow-up and returned two years later with recurrence of a markedly aggravated vascular epulis, prompting referral to the Maxillofacial Surgery and Medical Genetics departments, of the Ibn Rochd Hospital in Casablanca, for comprehensive evaluation and planning of complete excision under general anesthesia, considering the hemorrhagic risk, the extent of the lesions (**Figure 7**).

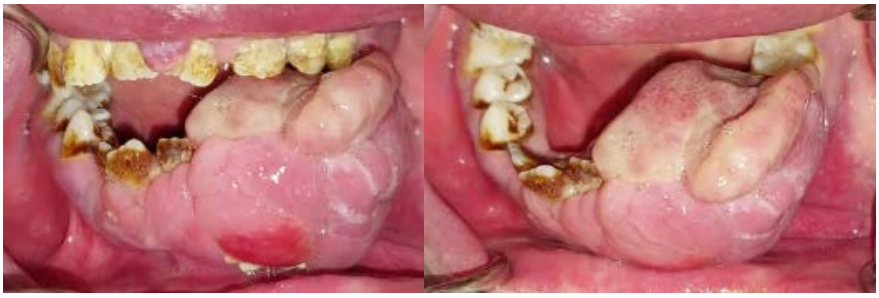

Figure 7. Recurrence of gingival vascular epulis after two years.

3. Discussion

SWS, also known as encephalotrigeminal angiomatosis, is a rare congenital neurocutaneous disorder characterized by vascular malformations involving the skin, brain, and eyes. It arises sporadically and is not inherited, resulting from a somatic activating mutation in the *GNAQ* gene. Classified among the phacomatoses, this condition typically presents at birth and has an estimated prevalence of approximately 1 in 50,000 live births [4].

The physiopathology of SWS involves a somatic *GNAQ* mutation. This mutation happens during the early stages of cerebral vascular development, when the blood supply to the brain, meninges, and facial structures is undergoing reorganization. This leads to progressive dilation of capillaries within the primitive vascular plexus. Consequently, the overlying leptomeninges become abnormally hypervascularized, while the underlying cortex progressively develops atrophy and characteristic calcifications [1] [5].

The clinical presentation of SWS is heterogeneous and typically encompasses dermatological, neurological, ophthalmologic, and oral manifestations [4].

Dermatological signs include a port-wine stain (capillary malformation), which represents the most common manifestation and is present in 87% to 90% of cases. It typically involves the face following the distribution of the trigeminal nerve, and may sometimes extend to the neck, chest, limbs, and back. The lesion is usually unilateral on the right side, although 33% of cases present with bilateral involvement. Its appearance tends to evolve with age, shifting from a lighter coloration

to a darker one [4] [6].

Neurological signs include leptomeningeal angiomatosis, which leads to contralateral hypertrophy and progressive cerebral calcification. Epileptic seizures typically appear during the first year of life, and intellectual disability may also be observed [1] [4].

Ophthalmologic signs in SWS include angiomas following the distribution of the trigeminal nerve, particularly its ophthalmic division, leading to visual acuity disturbances and a high prevalence of glaucoma [1] [5]. In our case, the patient presented with right-eye glaucoma and left eye strabismus, the latter being associated with right cerebral atrophy demonstrated on brain MRI.

One of the earliest clinical classifications of SWS, the Roach Scale, categorizes encephalofacial angiomatosis into three types (**Table 1**): Type I, characterized by both facial and leptomeningeal vascular malformations and often associated with glaucoma (the classic form of SWS); Type II, involving isolated facial capillary malformations with possible glaucoma; and Type III, presenting with isolated leptomeningeal angiomatosis, typically without glaucoma [2]. Our case corresponds to Type I according to the Roach Scale, as the patient presented both facial and leptomeningeal vascular malformations, consistent with the classic form of SWS. However, this classification has limited practical value, as treatment decisions must be made individually for each patient [1].

Table 1. The key manifestations of each SWS type according to the Roach Scale classification.

	Dermatological signs	Neurological signs	Ophthalmologic signs
Type I	+	+	+/−
Type II	+	−	+
Type III	−	+	−

SWS could include oral manifestations in 38% of the patients. When they occur, they typically follow the distribution of the trigeminal nerve, particularly its maxillary and mandibular divisions. As a result, patients may present with unilateral hemangiomas involving the gingiva, lips, tongue, palate, and floor of the mouth [7]. The clinical features of these manifestations may be further complicated by the use of antiepileptic medications, such as phenytoin or valproic acid, which are well known to induce gingival overgrowth. Additionally, intellectual disability can hinder effective oral hygiene, exacerbating the severity and recurrence of oral lesions [1] [5].

In the present case, a biopsy-excision of the gingival enlargement was performed before the initiation of antiepileptic therapy. Histological examination confirmed a vascular epulis, a benign gingival lesion likely potentiated by the generalized hypervascularization characteristic of SWS [2]. This finding ruled out drug-induced gingival overgrowth at that stage. Although lobular capillary hemangioma (pyogenic granuloma) has been reported in patients with SWS, the his-

topathological features observed in our case did not support this diagnosis and were instead consistent with a nonspecific vascular epulis [2].

Two years after the patient began valproic acid therapy, the gingival enlargement recurred, presenting as a more extensive and pronounced overgrowth. This recurrence was likely multifactorial, influenced by suboptimal oral hygiene and by the known gingival effects of valproic acid. These observations align with previous reports highlighting the high risk of recurrence of vascular oral lesions, particularly when oral hygiene is compromised and antiepileptic medications are introduced [1] [5].

Patients with SWS present an increased risk of oral mucosal bleeding during routine dental procedures due to intra-oral angiomatosis, which is often compounded by soft tissue overgrowth and gingival hyperplasia [2] [3]. Hence, patient safety must take precedence over the extraction of teeth surrounded by angiomatous tissue. A thorough pre-operative evaluation is therefore crucial, with careful anticipation of possible complications linked to the vascular characteristic of SWS. Several measures may be required to control or reduce hemorrhagic risk, including preoperative blood typing and cross-matching, ensuring the availability of transfusion support, the use of local hemostatic agents, or adjunctive techniques such as sclerosing injections or percutaneous transcatheter vascular embolization using materials like gel foam or polyvinyl alcohol. For these reasons, dental interventions should ideally be undertaken by a multidisciplinary team within a specialized pediatric dentistry or maxillofacial surgery unit, where appropriate expertise and emergency resources are readily available [2] [3]. In our case, management within a hospital setting, with a diode laser readily available, even though it was ultimately not required, combined with the use of hemostatic sponges and sutures, was sufficient to achieve hemostasis.

The differential diagnosis of SWS relies primarily on a combination of clinical examination and radiological findings. Key features include the characteristic unilateral port-wine stain, leptomeningeal angiomas, and ocular involvement such as glaucoma, which help distinguish SWS from other syndromes with vascular lesions, including Klippel Trenaunay Weber syndrome, neurofibromatosis, Bannayan Riley Ruvalcaba syndrome, and Coats disease [2]. Diagnosis is generally clinical and imaging based, and genetic testing for somatic GNAQ mutations is not routinely required, being reserved for atypical or research cases [2]. In our patient, the diagnosis of SWS was initially unrecognized, and it was the presence of oral manifestations specifically the vascular gingival overgrowth that prompted consultation in our oral surgery department. Collaborative evaluation with dermatology, neurology, and ophthalmology confirmed the syndrome, highlighting how dentists can play a pivotal role in the early detection of systemic diseases by recognizing local manifestations.

4. Conclusion

This case highlights the importance of oral manifestations as a potential first sign

of systemic disorders such as SWS. Vascular gingival overgrowth in SWS may serve as an early indicator, prompting multidisciplinary evaluation and appropriate management. Dentists, by recognizing these local signs, play a crucial role in the early detection and referral of patients with systemic vascular syndromes, emphasizing the value of thorough oral examinations in identifying underlying medical conditions.

Conflicts of Interest

The authors declare no conflicts of interest.

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