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Research Article

Periodontal Manifestations and Unusual Radiographic Features in a Patient with Sturge-Weber Syndrome: A Case Report

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Abstract

The Sturge-Weber syndrome or encephalotrigeminal angiomatosis is a rare neurological and congenital disorder with a frequency of 1 in 50,000 births. This syndrome is a nonhereditary developmental condition and is characterized by the presence of congenital capillary malformation and a hamartomatous vascular proliferation involving the face (port-wine stain or facial birthmark), sometimes skull and the tissues of brain, jaws, oral soft and hard tissues and rarely other body organs. Seizures, mental retardation, and cortical calcification (tram-tracks) and congenital glaucoma may be seen in this syndrome. We report here a 40-year-old female with Sturge-Weber syndrome associated with bilateral cutaneous capillary malformation on her face, neck, hands and feet and also gingival enlargement.

Key words: **Sturge-Weber syndrome, port-wine stain, angiomatosis, hamartoma.**

Introduction

The Sturge-Weber syndrome or encephalotrigeminal angiomatosis is a rare neurological and congenital disorder. Its incidence and the exact cause is unknown although evidence indicates that this syndrome has an embryologic origin and a genetic basis; an incomplete penetrance has been suggested for this disease.[1–4] This syndrome was initially described by Rudolf Schirmer in 1860 and further explained by Weber in 1879.[5,6] It is a nonhereditary developmental condition characterized by a congenital capillary malformation and hamartomatous vascular proliferation involving the face (port-wine stain or facial birthmark), skull and the tissues of brain, oral soft tissue as well as alveolar bone hypertrophy and rarely other body organs.[1-3,7] Congenital glaucoma, and cortical calcification (tram-tracks) may also be seen in this syndrome.[8-10] It is complicated when associated with other clinical features like neurologic deficits, including seizures, contralateral hemiparesis, hemiatrophy, headache, transient stroke and behavioral problems.[3,11] The Sturge-Weber syndrome is not associated with intracranial neoplasms and its typical triad features are: (1) facial

angioma; (2) the leptomeningeal angioma, particularly involving the occipital and posterior parietal lobes; and (3) choroidal angioma.[11,12] The facial stain can vary in color from light pink to deep purple and is caused by an overabundance of capillaries (tiny blood vessels) around the trigeminal nerve just beneath the surface of the involved skin. This syndrome occurs in about one in 50,000 newborns while capillary malformation occurs in 0.1−2% of newborns. Sturge-Weber syndrome affects both sexes equally and all races[13,14] and at present, unfortunately, no preventative measures are available against the disease and it cannot be diagnosed before birth. The disease is incurable but it is not fatal.[15]

Sturge-Weber syndrome is especially important for oral and maxillofacial surgeons and periodontists because of the involvement of the head, face and the oral cavity and also because of the serious complications that may arise from it. Intraoral manifestations can be readily recognized on physical examination. However, bony lesions are difficult to identify and may profoundly aggravate the prognosis of dental problems.[16]

Oral and periodontal manifestations of the disease occur in about 40% of patients.[17] In intraoral examination of most patients a significant and generalized hypertrophy of gingival tissues and dental anomalies can be seen. Also, significant histological and morphological changes of gingiva, periodontium and pulp have been observed. Lesions vary in color from light pink to deep purple or dark-red lesions and can be found anywhere in the mouth and blanch on pressure (positive diascopy). Lesions usually involve the maxilla, mandible, and floor of the mouth, lips, buccal mucosa, palate (hard and soft) and tongue. However, in the literature the most common feature is a gingival hyperplasia (vascular type).[1,3,13,15] In this report, we present a case of Sturge-Weber syndrome with oral and periodontal manifestations and an unusual radioghraphic appearance.

Case description

A 40-year-old woman with bilateral purplish cutaneous capillary malformation on the face, neck, hands and feet, was referred to the Department of Periodontics, Faculty of Dentistry, Tabriz University of Medical Sciences. Her chief complaints were severe gingival enlargement, gingival bleeding, suppuration exuding from the gingival tissues and halitosis. Extraoral examination revealed bilateral distribution of port-wine stains along the multiple segments of trigeminal nerve (the first, second, and third divisions) and on both sides of the forehead, cheeks, and upper eyelid. They were also found on the medial part of the nose and upper cutaneous lip with asymmetry and swelling on the lower lip.

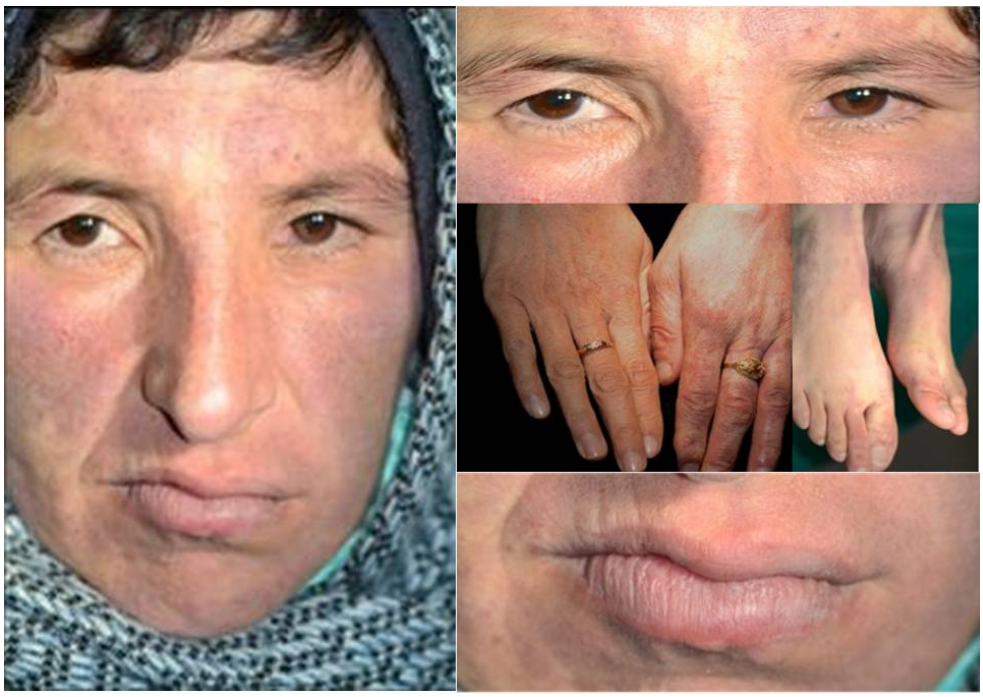


Figure 1. Photograph showing asymmetry of the face and bilateral facial port-wine stain, involving the medial aspect of the cheek, upper and lower cutaneous lip and nose. Not the Unilateral swelling of the lower lip and Port-wine stain that covers the hands and feet.

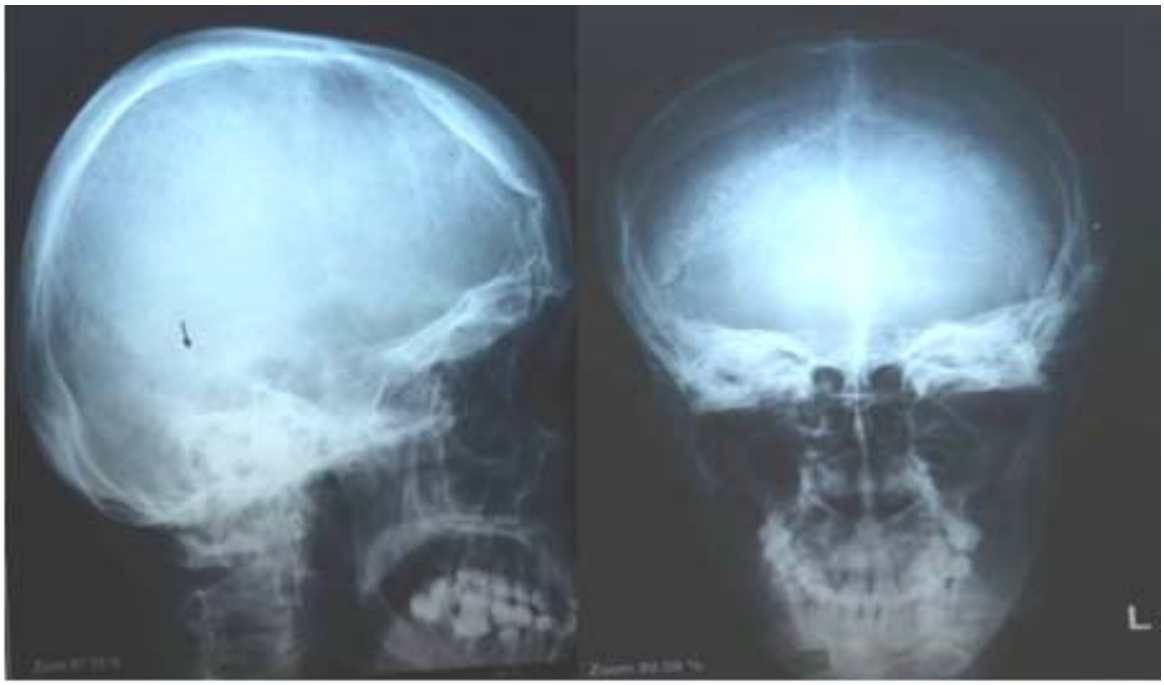


Figure2. Extraoral radiograph showing port-wine stain along ophthalmic branch of the trigeminal nerve and dilation of ocular blood vessels in the left eye.

There were cutaneous, vascular and port-wine lesions on the chest, abdomen and back and as well as on the head, the medial aspect of the hands and feet. The left side of her body was affected more severely than the right side (Figures 1). These lesions were present since birth and had gradually enlarged.

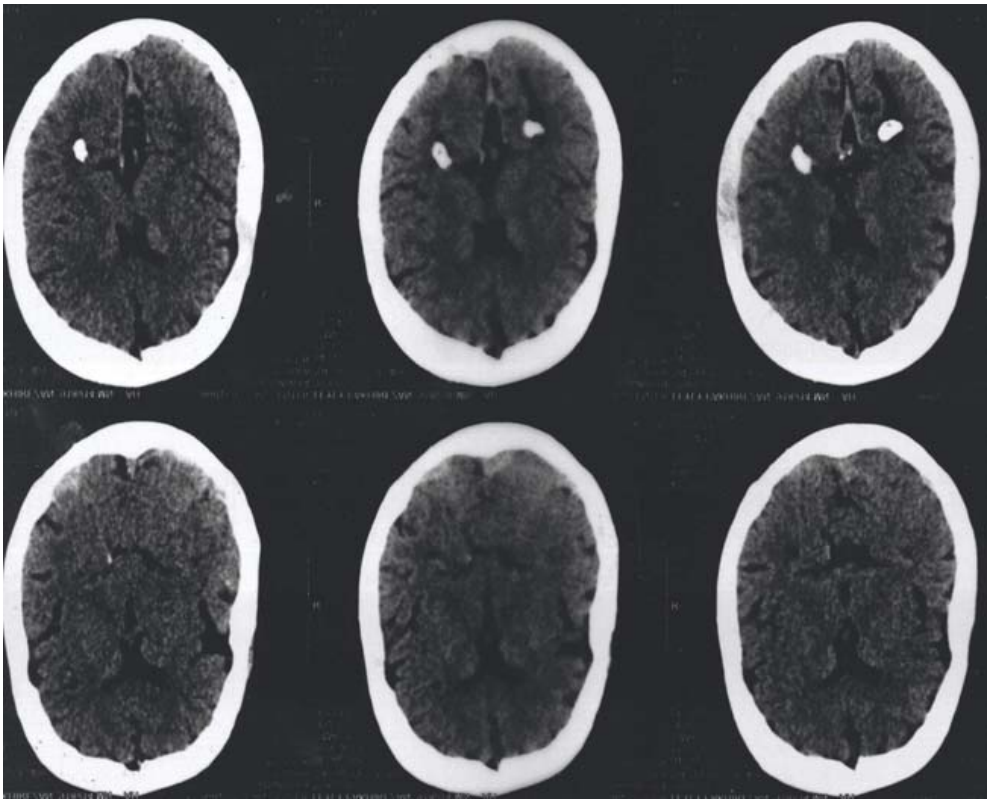


Figure 3. Cranial computed tomography did not reveal cortical and sub-cortical gyriform calcifications of the brain with cortical atrophy.

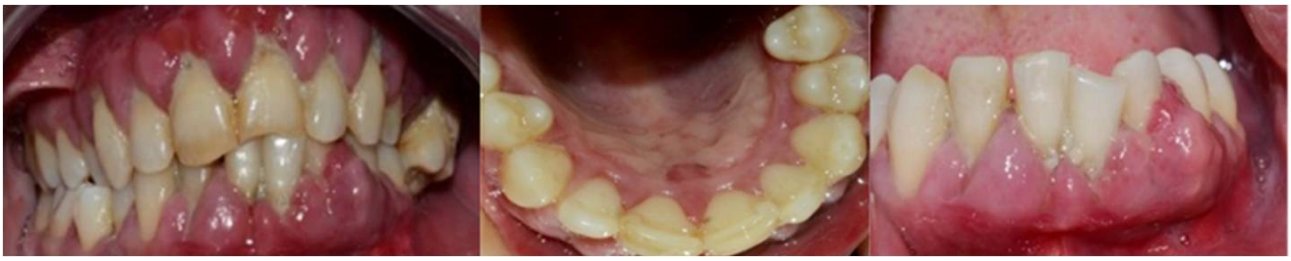
A comprehensive medical history was taken and physical examinations were carried out. She was mentally retarded in association with various behavioral problems and had been hospitalized for numerous stroke-like episodes and seizures and glaucoma of the left eye. She was taking phenytoin for many years due to her seizures, but she had not taken any anti-epileptic drugs in the last ten years. Her left eye was completely blind and she could only see shadows of things by her right eye due to glaucoma (Figure 2).

However, in this patient cortical calcifications, tram-line calcifications and cortical atrophy was not detectable by plain radiography and cranial computerized tomography (CT scan) (Figures 3).

Intraoral and periodontal involvement

Comprehensive intraoral examination revealed bulbous, fibrotic pedunculated and sessile diffuse and massive gingival enlargement on both sides particularly in the midline and left side of the maxilla and mandible. Both buccal and lingual gingivae were involved. The ability for self-cleansing for gingival hypertrophy was not possible and therefore the patient's oral hygiene was poor. Gingival hypertrophy had created periodontal pseudopockets giving a clinical picture of combined enlargement (fibrous and inflammatory). Deep periodontal pocket and trapping dental plaque had resulted in chronic periodontitis, halitosis, suppuration, and mobilization of teeth. There was a history of spontaneous gingival bleeding and bleeding on brushing (Figure 4).

Intraoral periapical radiographs showed coarse bony trabeculae and enlarged marrow spaces, PDL widening and spacing between teeth. Alveolar bone loss was revealed on the mandible central and lateral



Figures 4. Intraoral photograph shows generalized gingival hyperplasia and the extent of the lesion on buccal and palatal surfaces, as well as in anterior mandible.

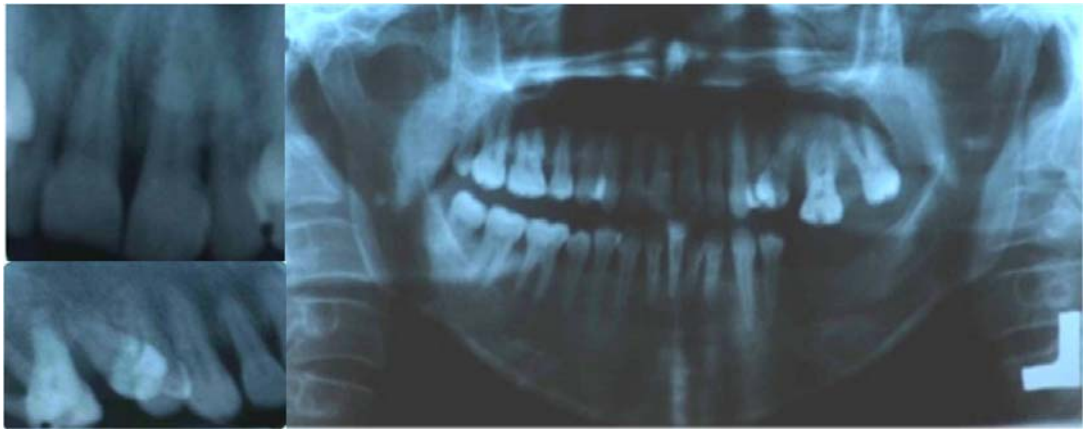


Figure 5. A panoramic and Intraoral periapical radiographs revealed malpositioning of teeth.

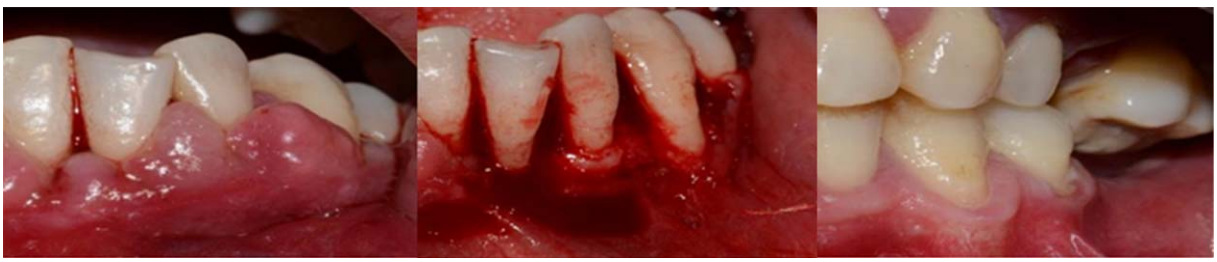


Figure 6. Intraoral photograph after scaling and root planing. Incisional biopsy and post-operative intraoral photograph of mandibular anterior portion showed regression of buccal lesion after surgery.

incisor regions. Widening of periodontal ligament space was seen in lateral incisors and first and second premolars in both maxillary and mandibular arches (Figure 5). Panoramic radiograph showed malpositioning of teeth (Figure 5).

Gingival hypertrophy was treated by periodontal surgery to remove the excess tissues and to allow for thorough debridement after complete scaling and root planing. Incisional biopsy of gingival overgrowths was performed (Figure 6) with safety margins during periodontal surgery procedure and histopathology feature showed a hyperplastic dense fibrotic connective tissue covered by parakeratotic stratified squamous epithelium which led to the diagnosis of focal fibrous hyperplasia with chronic periodontitis.

Discussion

Sturge-Weber syndrome (SWS) is a rare congenital disorder and usually sporadic. It is defined by the development of facial capillary malformation (port-wine stain) associated with specific ocular and neurologic abnormalities or leptomeningeal angioma.[15,18,19] The exact etiology of SWS is unclear. Some researchers believe it is caused by the persistence of the transitory primordial sinusoidal plexus stage of blood vessel development, and others suggest that etiology of SWS is capillary malformations resulting from somatic mutations in fetal ectodermal tissues.[20–22] Present evidence indicates that this syndrome has an embryologic and a genetic basis with incomplete penetrance.[1-4] In spite of being uncommon, this syndrome represents the most frequent disease among the neurocutaneous syndromes.[23]

The syndrome is frequently defined by the presence of at least three atypical signs of nevus flammeus, numerous stroke-like episodes and seizures and glaucoma. The condition has a progressive nature and neurologic deterioration may occur.[19,24-26]

Malformed cortical vessels in Sturge-Weber syndrome have been reported to be innervated only by noradrenergic sympathetic nerve fibers.[27] Most patients (80-87%) have epilepsy which may develop at any age, although they usually start in early childhood, and more than 50% have mental deficiency.[28-30] Developmental delay and mental retardation may be seen in 50−75% and emotional/behavioral problems in 85% of patients. Usually, these patients need special education in relation to seizures in adults.[31]

Sturge-Weber syndrome rarely occurs without the port-wine stain and its diagnosis is frequently defined by nevus flammeus in the face associated with convulsive attacks; of course, these symptoms are not its pathognomonic and they are not seen in all the patients.[17,23]

In the literature, various radiography examinations for Sturge-Weber syndrome have been suggested, including plain skull radiography (antero-posterior skull and lateral view), computed tomography scanning, magnetic resonance imaging, angiography, and at present nuclear medicine studies. Of course, the radiologic diagnosis of SWS is usually made with a high index of clinical suspicion and abnormal radiographic findings may be detectable only in 63% of patients.[2,9,10,30] This matter can result in complications in identification of this disease in cases with incomplete radiological but clinical manifestations.

Although cranial hemangiomas often show marked calcification of the brain blood vessels,[17,23] calcification and significant abnormalities were not detectable by plain radiography and cranial computerized tomography (CT scan) in the present case.

It must be pointed out that this patient suffered from numerous uncontrolled stroke-like episodes and seizures. She had glaucoma in her left eye for many years and since the patient had not been treated, her left eye was completely blind. Patients with a normal CT scan but an abnormal mental status need more thorough evaluations which might furnish guiding clues to pursue further investigations.

Although the radiologic findings (periapical and panoramic) in patients with Sturge-Weber syndrome are not specific, hamartomatous lesions may appear as well-circumscribed radiolucent lesions with sclerotic borders; in addition, bone erosion, coarse bony trabeculae and enlarged marrow spaces or cyst-like radiolucent lesions might be seen. These lesions may have a honeycomb or soap bubble appearance. Tooth displacement or root resorption of the teeth in the proximity of the lesion may occur.[33]

The treatment of the Surge-Weber syndrome depends on the pattern or intensity of its possible clinical manifestations.[32] Its treatment includes control of epileptic seizures and prevention of glaucoma and headaches. The port-wine nevus on the face usually improves by the use of new techniques of laser therapy or through the use of cosmetics.[34–36]

At intraoral examination of this patient, we found hemangioma lesions on the lips, palate, ventral and lateral mucosa of the tongue and the floor of the mouth,Also we observed generalized gingival hyperplasia almost on the entire buccal and lingual surfaces with malpositioning of teeth that require general management.

Hyperplastic lesions resembling tumor-like mass involved the papillary, marginal and attached gingiva of all the maxilla and mandible, especially the left side, causing spontaneous bleeding. Gingival hypertrophy had created periodontal pseudopockets giving a clinical picture of combined enlargement (fibrous and inflammatory). Because the patient had not taken any anti-epileptic drugs in the last ten years, gingival enlargement was probably due to plaque-induced inflammatory changes in the gingival tissues and chronic periodontitis that complicated mental retardation and disability in oral hygiene procedures.

In the present case, prior to treatment physician's permission was obtained and then we treated hypertrophic gingival tissue by gingivectomy and periodontal surgery to remove excess tissues and to allow for thorough debridement after complete scaling and root planing.

Of course, surgical excision with the Nd:Yag laser has been suggested in the literature for optimal management of hemorrhage.[13] There was normal bleeding during surgical treatment for gingival overgrowth in this patient.

Conclusion

The different signs and symptoms of Sturge-Weber syndrome have been described in the literatures. The syndrome has been referred to as complete when there are at least three cutaneous, ocular, and neurological signs (complete or classical form) and incomplete when only two areas are affected without the other. In the present case, Sturge-Weber syndrome was complete because central nervous system anomalies (numerous uncontrolled stroke-like episodes and seizures and mental retardation), glaucoma in her left eye and facial angiomas were present. This syndrome is extremely important to periodontists and oral and maxillofacial surgeons because it has a high frequency rate of vascular proliferation on the face and the oral cavity. Therefore, their knowledge about this syndrome and its complications for the successful management of these patients is necessary. Treatment of hypertrophic gingival tissue may be challenging due to the risk of hemorrhage and uncontrolled stroke-like episodes. To reduce the risk of hemorrhage, consultation with a physician, periodic systemic evaluation and meticulous oral examination are critical.

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