Case Report

Sturge-Weber syndrome – an unusual presentation

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Abstract

Sturge-Weber syndrome (SWS), sometimes referred to as encephalotrigeminal angiomatosis, is a rare congenital neurological and skin disorder of unknown etiology. The classic pathognomonic features of disease include angioma of the leptomeninges extending to cerebral cortex with ipsilateral angiomatous lesions, unilateral facial nevus affecting the division of trigeminal nerve and epileptic convulsions. The most characteristic oral manifestation is represented by gingival hemangiomatous lesion usually restricted to ipsilateral maxilla or mandible. A classic case of Sturge-Weber syndrome with extended involvement of other parts of the body is reported here.

Key words: Encephalotrigeminal angiomatosis; Port wine stains; Sturge-Weber syndrome; Tram line calcifications

Introduction

Sturge-Weber syndrome (SWS), sometimes referred to as encephalotrigeminal angiomatosis, is a rare congenital neurological and skin disorder (1). It was first described by Schirmer in 1860 and later more specifically by Sturge in 1879, who 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 (2).
Sturge-Weber syndrome

It is a rare disorder occurring with a frequency of 1:50,000 live births (3). Both sexes are affected equally and no racial predilection is seen (4). The classic feature of this disorder is the angioma of the leptomeninges. Most common features are epilepsy, Port-wine stain, dermal angiomas, and abnormal findings in skull radiographs, mental retardation, ocular involvement and hemiplegia (5). Oral manifestations of the disease may vary considerably. However the most common feature is a gingival hemangiomatous lesion usually restricted to ipsilateral maxilla, mandible, floor of the mouth, lips, cheeks, palate and tongue (1).

In this report, we present a case of SWS who presented with its characteristic manifestations along with extended involvement of the other parts of the body.

Case report
A 32 years old female patient born to non-consanguineous married parents reported with a chief complaint of bleeding from the gums since last 4 months. Past medical history revealed that the patient had developed convulsive disorder at the age of 6 months for which she was under medication. Her parents also gave history of delayed developmental milestones and learning disabilities. History of hysterectomy done 10 years back due to excessive bleeding was also revealed.

Extra oral examination showed presence of port-wine stain on left side of face along the ophthalmic and maxillary ramifications of trigeminal nerve (Figure 1). Ipsilateral labial angioma was also detected. These port-wine stains were seen extending over the left side of the neck, left hand, over the abdomen and then crossing the midline and extending over the right leg (Figures 2, 3). Examination of eyes revealed decreased vision and photophobia with respect to the left eye.

Intraoral examination showed angiomas in the gingival mucosa, buccal mucosa, lip and palate on the affected site and also gingival hyperplasia (Figures 4, 5). The lesions showed positive diascopy. Patient had poor

Figure 1: Extra-oral photograph showing port-wine stains of the left side of the face

Figure 2: Photograph showing angiomatous involvement of left neck and left hand

Figure 3: Photograph showing involvement of the right leg
oral hygiene. Tomographic examination was carried out and showed the presence of thin meningeal gyral calcifications in the occipital lobe (Figure 6). Based on the history, clinical and radiological findings a diagnosis of Sturge-Weber Syndrome was made.

**Figure 4:** Intra-oral photograph showing angiomatous involvement of the left lip, buccal mucosa and palate

**Figure 5:** Intra-oral photograph showing angiomas of the gingival with hyperplasia

Patient was advised a thorough plaque control regimen. It included oral prophylaxis, use of chlorhexidine mouth rinses and oral hygiene instructions. A considerable decrease in gingival bleeding on chewing food was noticed after the plaque control therapy. Patient was advised a periodic follow up. Furthermore, ocular and neurological examinations were advised.

**Figure 6:** CT images showing intracranial calcifications

**Discussion**

SWS is an embryonic developmental anomaly resulting from errors in mesodermal and ectodermal development. Unlike other neurocutaneous disorders (phakomatoses), Sturge-Weber occurs sporadically (i.e., does not have a hereditary etiology (6). The clinical diagnosis of SWS is usually definitive; however, the recognition of this disease may be complicated in case of incomplete radiological and clinical manifestations. The encephalotrigeminal angiomatosis has been classified in three types: Type I, which is characterized by facial and leptomeningeal angiomas and possibly glaucoma (classic syndrome); Type II in which facial angiomas are detected without neurological disturbances; Type III that is represented by the presence of isolated leptomeningeal angioma (7).

In the present case, there was presence of facial angiomas with meningeal calcifications. Patient exhibited neurological involvement such as seizure and also ocular alterations. Hence the patient was fit in the type I of SWS.

The main differential diagnosis of SWS includes Klippel-Trenaunay-Weber syndrome and Beckwith-Wiedemann syndrome. Klippel-Trenaunay-Weber syndrome consists of port-wine stains of the extremities and face with hemi hypertrophy.
Sturge-Weber syndrome

of soft and bony tissues (8) which was absent in the present case. Beckwith-Wiedemann syndrome consists of a facial port-wine stain, macroglossia, omphalocele, and visceral hyperplasia. A risk of visceral neoplasia is also noted (9). It was also missing in the present case and helped in differentiating it from these two.

In general, the skin findings are considered extremely important for the early diagnosis of neurocutaneous disorders. Nevertheless, the absence of facial angiomas does not exclude the diagnosis of SWS (10). Aydin et al., (7) reported a case of a patient without facial angiomas but other features of the SWS were detected such as seizures and leptomeningeal angiomas. The literature has demonstrated that the distribution of the facial angiomas is associated to the trigeminal nerve ramifications (11, 12). However, according to Inan and Marcus (10) this pattern can vary considerably and does not follow exclusively the vascular or dermatological pattern since additional lesions may occur on the palms, soles and other parts of the body (13). In addition, the port wine nevi are localized in the face, especially on the right side, and are detected in 87 to 90% of the cases (10). The lesion extension over the middle line is observed in 50% of the patients and bilateral involvement can be detected in about 33% of the cases. In the present case, port-wine stains were found on the left side of the face involving the ophthalmic and maxillary ramifications of the trigeminal nerve. And along with this, there was an unusual presentation of extensive involvement over the neck, left hand, abdomen and then crossing the midline and extending over the right leg.

Characteristically leptomeningeal angiomas occur as unilateral lesions affecting the pia-arachnoid membrane over the posterior temporal, parietal and occipital areas (2). It commonly shows abnormal blood flow pattern as venous occlusion, thrombosis, vasomotor phenomenon and vascular steal phenomenon resulting in cortical ischemia. This in turn gives rise to epileptic convulsive crisis, transient hemiparesis, gliosis and progressive deposition of calcium salts. These calcifications produce a characteristic double contoured “tram-line” appearance following the convolutions of cerebral cortex. Brushfield and Wyatt stated that these tram-line calcifications are pathognomonic of SWS (13). These calcifications appear after the patient reaches 2 years of age and remain stationary after second decade of life. These calcifications are gyriform and curvilinear and most commonly seen in parietal and occipital lobes as seen in our case.

In regard to the oral findings, hyper vascular changes are detected (11, 12). These alterations are basically related to angiomas at the gingival mucosa, lip and palate basically on the affected side and also gingival hyperplasia. It is characterized by increase in the vascular component and gingival hemorrhage at minimal traumatisms. The oral manifestations are generally unilateral and finish abruptly in the midline. Macroglossia and maxillary bone hypertrophy found in some patients can cause malocclusion and facial asymmetry. The gingival hyperplasia in these patients could be secondary to anticonvulsant therapy further complicated by poor oral hygiene secondary to mental retardation (14).

In the present case also unilateral angiomatosus involvement of the buccal mucosa, lip, palate, gingiva with hyperplasia was noticed. The oral rehabilitation of patients with SWS is usually complex requiring conservative procedures and, in some cases, surgical approaches through an accurate planning and the utilization of techniques which diminish the bleeding. The present case was subjected to thorough plaque control regimen that helped in controlling the bleeding from the gingiva. The patient was
kept under observation and called for a periodic follow up.

Conclusion
Sturge-Weber syndrome is a rare phacomatoses and presents with large spectrum of clinical manifestations. Hyper vascular changes are detected in the oral cavity that leads to the bleeding. Bleeding from the oral cavity is further complicated due to poor oral hygiene secondary to the mental retardation and to certain extent due to the intake of anti-convulsive drugs. Since the etiology is not known, the disease cannot be prevented. Hence early diagnosis plays a very important role in preventing the future complications related to Central nervous system and oral cavity.

References