

## CASE REPORT

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### STURGE WEBER SYNDROME WITH UNUSUAL INTRACRANIAL FINDINGS: A CASE REPORT

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**ABSTRACT:** The Sturge-Weber Syndrome, also known as encephalotrigeminal angiomatosis, is a rare vascular neurocutaneous alteration. The main clinical features of this syndrome are facial vascular cutaneous naevus, usually unilateral, which often follows the outline distribution of trigeminal nerve. We hereby are reporting a clinical case of Sturge-Weber Syndrome in a 16 year old female patient who presented with oral, cutaneous and ocular manifestations related to the syndrome.

**KEYWORDS:** Sturge-Weber Syndrome; Port-Wine Stain; Tram Line Calcifications.

**INTRODUCTION:** Sturge-Weber syndrome is a sporadic neurocutaneous disease characterized by facial port-wine stain, ocular abnormalities (glaucoma and choroidal hemangioma) and leptomeningeal angioma.<sup>1</sup>

Sturge Weber syndrome (SWS) 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.<sup>2</sup> It is rare disorder occurring with a frequency of 1:50, 000 live births.<sup>3</sup> Both sexes are affected equally and no racial predilection is seen.<sup>4</sup>

The classic feature of this disorder is the angioma of leptomeninges. Most common features are epilepsy, Port-wine stain and dermal angiomas, abnormal findings in skull radiographs, mental retardation, ocular involvement and hemiplegia.<sup>5</sup> Oral manifestations of the disease may vary considerably and changes in morphology and histology of gingiva, periodontium and pulp have been reported.

However the most common feature is a gingival hemangiomatous lesion usually restricted to ipsilateral maxilla, mandible, floor of mouth, lips, cheeks, palate and tongue.<sup>1</sup>

**CASE REPORT:** A 16 year old female presented to our department with chief complaints of headache and mild photophobia since one year and progressive loss of vision since two months.

She did not have any mental retardation or delayed milestones, her intelligence was normal and there was no history of epileptic seizures in the past.

On examination she had a port wine stain over forehead, both the upper eyelids, lower face, right side of arm, forearm and palms and there was facial asymmetry. (FIGURE 1)

On examination of oral cavity she had a port wine stain on hard palate and mucosa of right side along with gum hypertrophy. Dentition was normal and there was no malocclusion. (FIGURE 2)

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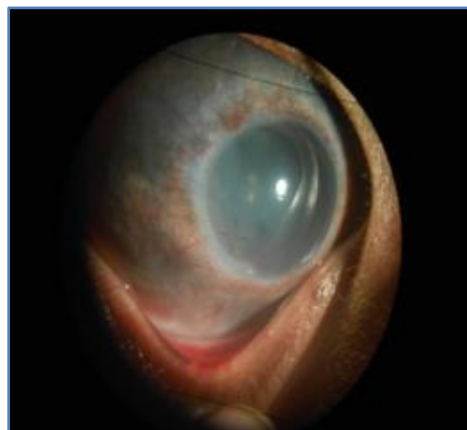
**Figure 1**



**Figure 2**



**Figure 3: Ocular examination revealed blue corneal sclera and circumpapillary congestion and bupthalmos in right eye**



**Figure 4**

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Fundus examination revealed hazy media in the right eye with cup: disc ratio of 0:9 with loss of neuroretinal rim. Left eye have cup: disc ratio of 0:7 with inferior notching. Slit lamp examination of right eye revealed glaucoma fleckers (FIGURE 4).

Her vitals were stable and her routine and biochemical findings were:

CBC revealed Hb-11.0 gm%, ESR- 15 mm, Total leucocyte count 8300 (N-69%, L-28%), platelet count - 2.3 lakh/mm<sup>3</sup>, blood urea - 12mg%, serum creatinine- 0.83mg%.

Her plain and contrast CT scan head revealed mild enlargement of pituitary possibly hyperplasia and diffuse enlargement of skull vault suggestive of fibrous dysplasia. Further MRI study of brain was done with MR angiogram neck vessel and intracranial circulation which further confirmed diffuse enlargement of skull vault with ground glass appearance suggestive of fibrous dysplasia and pituitary hyperplasia but there was no significant critical stenosis or aneurysm in intracranial vessels (FIGURE 5 & 6).

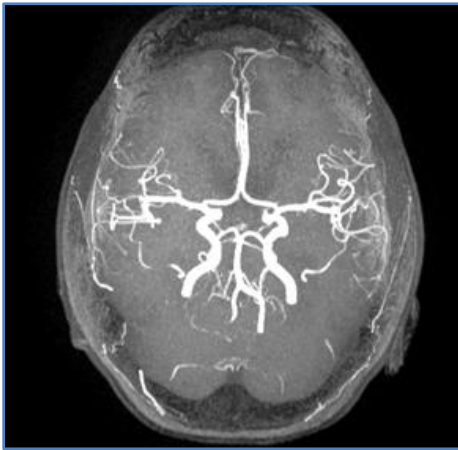


Figure 5

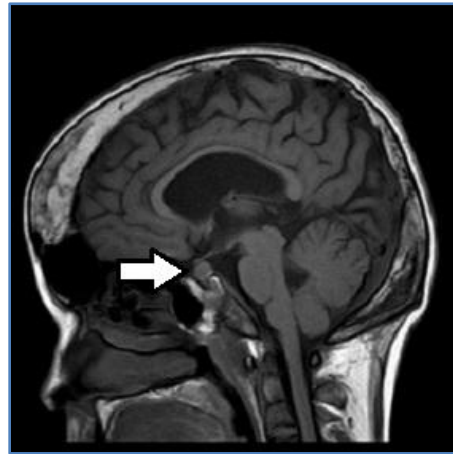


Figure 6

**DISCUSSION:** SWS is referred to as complete when both CNS and facial angiomas are present and incomplete when only one area is affected without the other.

The Roach Scale is used for classification, as follows <sup>1</sup>:

Type I - Both facial and leptomeningeal angiomas; may have glaucoma.

Type II - Facial angioma alone (no CNS involvement); may have glaucoma.

Type III - Isolated leptomeningeal angioma; usually no glaucoma.

According to the above criteria, our case is complete Type II SWS case. Neurological outcome in children with SWS is highly variable, ranging from minimal or no neurological signs to a devastating impairment with uncontrolled seizures, hemiparesis, visual field defect and progressive mental retardation.

Seizures is a very common feature, often occurs during the first year of life<sup>6</sup> and result from cortical irritability caused by angioma through the mechanism of hypoxia, ischemia and gliosis. About 80% of affected persons have focal seizures involving the contralateral side of the port wine stain<sup>7</sup> which was not found in our patient.

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The hypoperfusion of cortical tissue is further accelerated by seizures, thereby worsening the prognosis<sup>8</sup> Developmental delay and mental retardation are almost always associated with seizures while in our patient intelligence was normal and there was no mental retardation.

Neuroimaging studies help to establish the diagnosis, assess severity and follow the progression of brain involvement in SWS. Cortical atrophy underlying the angioma with gyriform "tram track" calcification is the characteristic imaging feature. Calcification however is unusual before 2 years of age and most commonly involves the parietal and occipital lobes. MRI is the current "gold standard" for diagnosis of disease which is reliable even in very young infants.<sup>9</sup>

The main ocular manifestations buphthalmos and glaucoma occur due to secondary increase in intraocular tension due to increased secretion of aqueous humor by choroidal hemangioma. Glaucoma and buphthalmos typically occur when port wine stain involves eyelids. It can develop at any age, is usually unilateral and ipsilateral to port wine stain. Other features include visual loss, macrocephaly and hemiatrophy similar to that in our patient.

The oral manifestations include Port-wine stain lesion of oral mucosa along with the hypervascular changes. Most common manifestation is angiomatous lesion of gingiva which can vary from slight vascular hyperplasia to massive hemangiomatous proliferation. It is characterized by increase in the vascular component and gingival hemorrhage at minimal traumatism.

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.<sup>1,3,10</sup>

Medical management of Sturge Weber Syndrome includes treatment for seizures, treatment for glaucoma with carbonic anhydrase inhibitors,  $\beta$ - antagonists to prevent optic nerve atrophy and use of aspirin for headache and to prevent vascular disease. Port wine stain requires treatment with pulsed tunable dye laser which should be started as soon as possible.

**CONCLUSION:** To conclude, the diagnosis and management of patients with Sturge - Weber syndrome requires the combined skills of a physician, radiologist and a psychologist and ophthalmologist. Seizures play a major role in failure of mental development and deterioration of mental function, hence effective seizure control is mandatory.

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