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**Case Report**

# Right sided facial nevus with contralateral cavernous angioma in a child with Sturge Weber syndrome

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## ABSTRACT

Sturge Weber syndrome is a rare sporadic neurocutaneous syndrome. It has extra -oral manifestations mostly in form of unilateral port wine stain on face that are ipsilateral to intra-cranial lesions (mostly lepto-meningeal angioma), seizures, glaucoma and intra-orally by hemangioma of ipsilateral gingiva. We case report a child with Sturge Weber syndrome with facial nevus opposite to intra-cranial lesion, intra-oral port wine stain and gum hypertrophy. Other than contralateral intracranial lesion and facial nevus, challenge was whether to see gingival manifestations as part of syndrome or as drug side effect, as child was also on phenytoin, an anti-epileptic drug widely related to this condition.

**Keywords:** Sturge-Weber syndrome, Contra-lateral facial nevus, Gingival hyperplasia, Phenytoin, Children

## INTRODUCTION

Sturge Weber syndrome (SWS), also known as Sturge-Weber-Dimitri syndrome or encephalotrigeminal angiomatosis is a rare congenital non-hereditary neurocutaneous syndrome characterized by unilateral facial cutaneous vascular malformations and ipsilateral leptomeningeal angiomatosis.[1] It results from neural crest dysgenesis in embryonic life. Vascular plexus develops around cephalic portion of neural tube around sixth week of intra-uterine life. This plexus undergoes regression during ninth week of intra-uterine life. Persistence of this vascular plexus is believed to cause Sturge Weber syndrome.[2]

This syndrome was first identified by Schirmer in 1860.[1] Detailed description of neurological, dermatological and ophthalmological manifestations was given by Sturge in 1879 and radiological changes of SWS were described by Weber in 1929.[3]

## CASE REPORT

8-year-old male child was brought to pediatric department OPD with primary complaint of poorly controlled seizures. A review of patient's medical history revealed that he was having seizures since 2 years of age. Seizures started as right side clonic seizure but became generalized after some months. He was on oral phenytoin since last two years only, started by some local practitioner. Prior to that he was not on any anti-epileptic drug. Child was product of non-consanguineous marriage, was born full term and had uneventful prenatal, natal and post-natal period. There was no mental retardation. Family history was not suggestive of similar complaints in his immediate and distant relatives. On examination, child had port wine stain on right side of face along the ophthalmic and maxillary division of trigeminal nerve. There was hemi hypertrophy of right side of face (Figure 1). He also had gum hypertrophy in upper jaw that was bilateral and not just limited to right half. Port wine stain was seen intra –orally also on right side soft palate (Figure 2). Rest of neurological examination was within normal limits. There was no focal neurological deficit, no cranial nerve palsy and no features of meningeal irritation. Eye examination was within normal limit.

On basis of clinical presentation and examination,

provisional diagnosis of Sturge-Weber syndrome was made and further investigations were done to confirm diagnosis. Blood investigations that included complete blood count, liver function test, kidney function test, coagulation profile were within normal limits. X-ray skull did not reveal any calcification. Magnetic resonance imaging (MRI) (contrast) of brain revealed cavernous angioma in left high frontal lobe, parafalcine in location. Both choroid plexus was bulky and hypertrophied with intense enhancement (Figure 3). There was no leptomeningeal enhancement. CECT brain and MRI brain further confirmed absence of any calcification. Eye examination by ophthalmologist revealed normal findings.

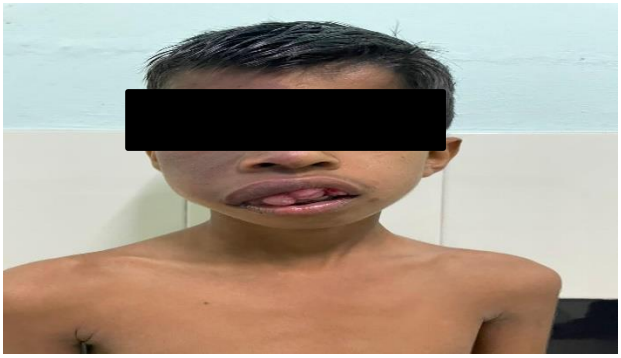

**Figure 1: Port wine stain on right-side of face along ophthalmic and maxillary division of trigeminal nerve, hemi hypertrophy of right-side face.**

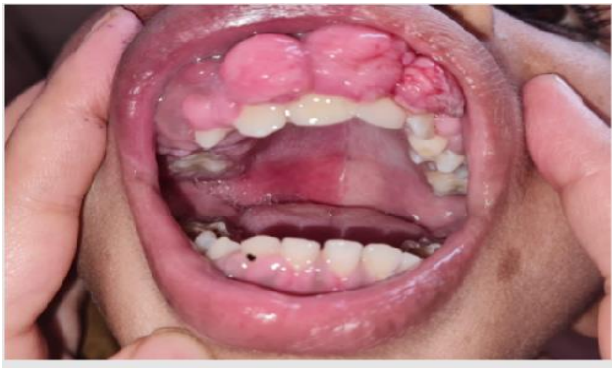

**Figure 2: Diffuse gum hypertrophy and intra-oral port wine stain on right side soft palate.**

Table 1 shows clinical manifestations of SWS and manifestations seen in our case.[2,3]

Gum hypertrophy was not limited to right side, was firm on palpation, non-blanchable, not prone to bleeding during probing and as per parents appeared after child was started on phenytoin. Hence, this finding was attributed to Phenytoin rather than gingival angiomatosis, a common intra-oral manifestation of Sturge-Weber syndrome. Oral anti-epileptic drug was shifted from phenytoin to levetiracetam.

Child responded well to oral levetiracetam, and in pediatric follow up seizures are under control. Child is also in follow up of neuro-surgery and dental department of our hospital.

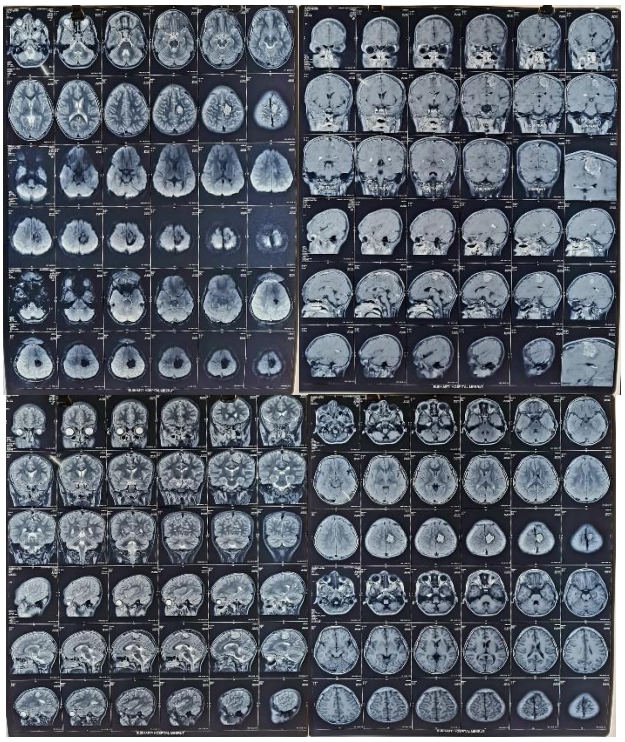

**Figure 3: MRI brain (contrast) showing cavernous angioma in left high frontal lobe. There is no lepto-meningeal enhancement.**

**Table 1: Clinical manifestations of SWS and manifestations seen in our case.**

| Clinical features | Incidence (%) | Present case |
|---|---|---|
| **Epilepsy** | 80 | + |
| **Port-wine stain** | 76 | + |
| **Abnormal radiographic findings** | 63 | + |
| **Mental retardation** | 54 | _ |
| **Oral manifestations** | 38 | + |
| **Hemiparesis** | 37 | _ |
| **Ocular manifestations** | 37 | _ |

## DISCUSSION

Sturge Weber syndrome is rare disorder occurring with frequency of 1: 50,000, without any gender and racial predilection.[4,5] Though its etiology is still poorly understood, studies have shown somatic mutation in the GNAQ gene located on long arm of chromosome 9 as a causal factor for SWS.[6] Sturge Weber syndrome is classified on basis of Roach scale, which is as follows: type 1 - both facial and leptomeningeal angiomas; may have glaucoma; type 2 - acial angioma alone (no CNS involvement), may have glaucoma; and type 3 - isolated leptomeningeal angioma; usually no glaucoma.[7]

Complete SWS implies presence of both CNS and facial angioma. However, if only one area is affected it is considered incomplete. In our case both facial and intracranial angioma was present, hence, according to above mentioned criteria it was a complete SWS type 1.

In SWS, areas of brain affected by angiomatosis have inadequately functioning superficial cortical venous system. This leads to central re-routing of blood through medullary veins causing venous stagnation and ischemia.[8] In ischemic areas, there is progressive deposition of calcium salts. Gyriform calcium deposit in cortex appears like" tram-track" in CT scan.

First manifestation of SWS is usually seizure, occurring in first year of life itself in 90% of cases. Seizure can occur in many forms like tonic, or clonic atonic and can be general or focal. It is considered that intracranial calcification are responsible for these epileptic foci.[9] Interestingly our case had seizure since second year of life but intra-cranial calcifications were absent.

SWS presents with ipsilateral facial nevus and intracranial lepto-meningeal angioma in 80% of cases. Few variants from this typical presentation have been well described in literature. These variations include bilateral facial angiomas, absent facial nevus.[10-12] However, facial nevus contralateral to intra-cranial lesion is very rare finding. We could find only two such cases in literature, that included one pediatric and one adult case.[13,14] Other interesting finding of our case was presence of localized cavernous angioma as intracranial lesion. Leptomeningeal enhancement/angioma which is most characteristic MRI finding of SWS was absent in our case. Absence of Leptomeningeal enhancement in SWS has been seen in rare cases only.[12] We also observed port-wine stain on soft palate ipsilateral to facial nevus.

Patient also had gum hypertrophy. As patient was on phenytoin, challenge here was to attribute this particular condition to phenytoin or consider it as part of SWS. Phenytoin is well known to cause gum hypertrophy as side effect, while gum hypertrophy due to gingival angiomatosis is a common intra-oral manifestation of SWS.[1] In our case, gum swelling was not restricted to one quadrant of jaw, it was firm to touch, non-blanchable, did not bleed on probing and appeared in last two years only after phenytoin was started. Hence gum hypertrophy was considered a side effect of Phenytoin in our case. Anti-epileptic was switched from phenytoin to levetiracetam and seizures were well controlled.

Treatment of SWS is mainly supportive. Anti-epileptics should be started soon after first seizure. Cases with refractory seizures are considered for surgical procedures, like hemispherectomy or focal resection of epileptic focus.[6] Other supportive treatments are dye-laser photocoagulation for Port –wine stain and treatment for increased intra-ocular pressure in glaucoma.[6]

Differential–diagnosis of SWS include Von Hippel–Lindau disease, Wyburg-Mason syndrome, Klippel–Trenaunay-Weber syndrome, and Blue-Rubber Bleb nevus syndrome.[6]

## CONCLUSION

SWS is a rare non hereditary genetic syndrome with no specific treatment. Here we presented a case with facial nevus and contra-lateral intracranial lesion, making it an extremely rare case. Leptomeningeal enhancement a characteristic finding of SWS was absent in our case, we also observed intra-oral port wine stain and gum hypertrophy due to phenytoin, which had to be differentiated from gingival hemangioma of SWS.

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