



## Sturge –Weber Syndrome – A Case Report

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### Abstract

*Sturge-Weber Syndrome or encephalotrigeminal angiomas is a rare congenital, non-familial disorder characterized by a congenital facial birthmark and neurological abnormalities. The GNAQ gene mutation is responsible for Sturge-Weber syndrome.*

### Introduction

Sturge–Weber syndrome (SWS) is also called encephalotrigeminal angiomas. It is a rare non-hereditary condition characterized by a facial cutaneous vascular nevus (nevus flammeus or port-wine stain) in association with leptomeningeal angiomas.<sup>1</sup> Schirmer first described it in 1860 in association with facial angioma and buphthalmos. Later in 1879 while reporting a case of 6½ year old girl, William Allen Sturge acclaimed these features in accordance with neurological findings. There is a risk of 10–50 % for the involvement of brain if a child is born with Port-wine birthmark (PWB) on the forehead or the upper eyelid. In 1992, Roach categorized SWS variants into three types:

- Type I: individual has a facial PWS, leptomeningeal angioma, and may have glaucoma
- Type II: individual has a facial PWS, no leptomeningeal angioma, and may have glaucoma

- Type III: individual has leptomeningeal angiomas, no facial PWS, and, rarely, glaucoma.

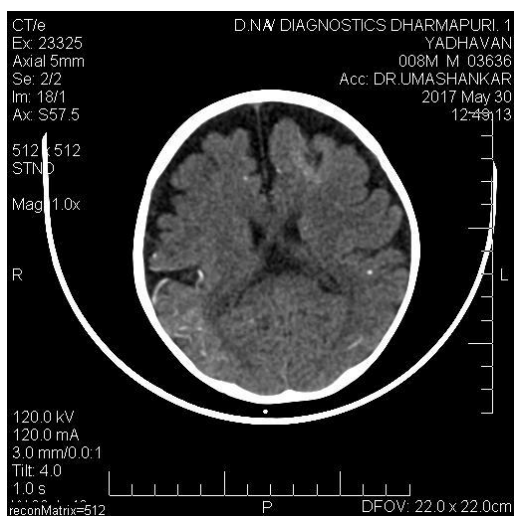
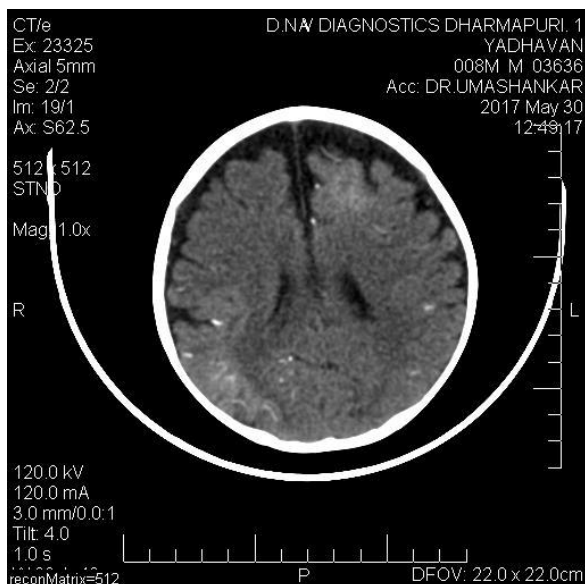
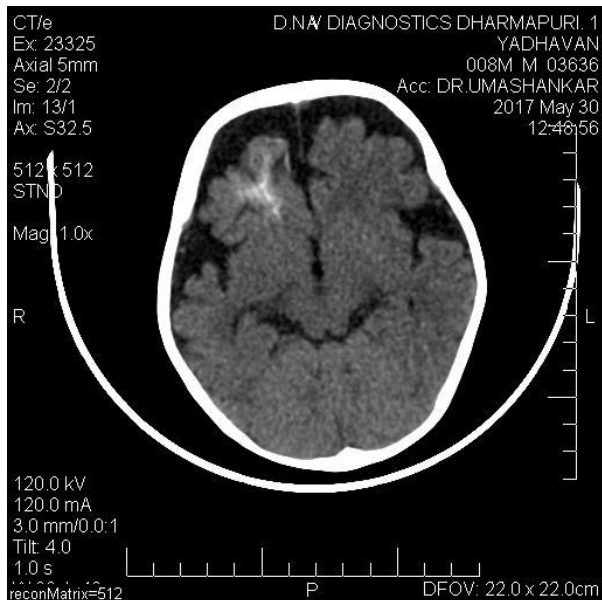
### Case Report

A 8 month old boy came to the center with complaints of seizures and reddish discoloration of the skin involving the right forehead and eye. While obtaining further history it was known that the reddish discoloration was present from birth and there has been no increase in size or change in color. The family history was non-contributory. It was a normal full term delivery without any complications.

CT of the brain showed scattered specks of calcifications in bilateral frontal and parietal regions.

Subtle gyral calcifications are seen in the left frontal and right high parietal regions.

Calcifications seen in the right basifrontal region  
Prominent vessels seen in the right temporal and bilateral parieto-occipital regions.



### Discussion

The Sturge-Weber Syndrome was initially described by Schirmer, in 1860, and was later specified by Sturge (1878), who associated the dermatological and ophthalmic changes to the disease's neurological manifestations. In 1992, Weber complemented it through the documentation of these patients cerebral radiologic alterations <sup>(2)</sup>. The inheritance of Sturge-Weber syndrome is sporadic and it occurs with a frequency of 1:50,000 <sup>(1)</sup>. Both the sexes are equally affected and no racial difference has been reported <sup>(4)</sup>. It is believed that it is caused by the abnormal persistence of an embryonal vascular system, which is localized around the cephalic region of the neural tube <sup>(3)</sup>. The SWS presents with neurological, cutaneous, ocular and oral manifestations that may (or not) be associated with one another; however the most evident clinical manifestation is the presence of the nevus flammeus or port wine stain on the face, which normally follows the course of branches V1 and V2 of the trigeminal nerve <sup>(2)</sup>. According to INAN (1999), the port wine nevus is localized on the face especially over the right side and are detected in 87 to 90% of the cases. 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 <sup>(5)</sup>. In our case, the patient showed nevus flammeus only on the right side of the forehead, upper eyelid and nose without extension over the middle line.

### Conclusion

The treatment of the Sturge–Weber's syndrome is variable and depends on the presentation or intensity of its possible clinical features. The wide spectrum of clinical manifestations of SWS leads to multidisciplinary approaches for its management, such as Neurophysician, Ophthalmologist, Cosmetologist, Physiotherapist, Radiologist and Dentist

### References

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