# Sturge Weber Syndrome with Absent Facial Nevus and Fronal Leptomeningeal Angioma: A Rare Dilemma

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**Abstract**

Sturge Weber Syndrome (SWS) is a rare, sporadic, neurocutaneous disorder characterized by cutaneous facial angioma, leptomeningeal angiomas and glaucoma. The incidence of SWS without facial nevus is unknown. However, it is rare with only few patients reported in the literature. The occipital lobe is characteristically involved in SWS .Other lobes are rarely involved especially the frontal lobe .The association between SWS without facial nevus and frontal lobe angiomas is extremely rare. In this study, we present 2 cases of this syndrome with absent facial and associated frontal leptomeningeal angiomas.

**Keywords:** Sturge Weber syndrome, Facial nevus, Leptomeningeal angioma, Frontal lobe.

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## Introduction

Sturge Weber Syndrome (SWS) is a rare, sporadic, neurocutaneous disorder characterized by cutaneous facial angioma, leptomeningeal angiomas and glaucoma[1)]. It is caused by somatic mosaic activating mutation in GNAQ[2)]. It is classified into three types: Type I (facial and leptomeningeal angioma with possible glaucoma), type II (facial angioma without evident intracranial involvement), and type III exclusive leptomeningeal angioma[3)].

The incidence of SWS without facial nevus (type III) is unknown. However, it is rare with only few patients reported in the literature[4-7)]. The occipital lobe is characteristically involved in SWS; however, temporal, parietal and frontal lobes are involved with decreasing frequency respectively[8)]. The association between SWS without facial nevus and frontal lobe angiomas is extremely rare. Here, we present two cases of this syndrome with absent facial and associated frontal leptomeningeal angiomas.

## Case Report

### Case 1

A twenty-month old boy presented with a history of febrile generalized tonic clonic

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convulsion was examined. Apart from mild motor delay, his general, ophthalmology and neurological examinations were normal. His EEG was normal. His brain computed tomography (CT scan) at 20 months of age showed subcortical calcification in the right frontal region with focal atrophy in the right frontal lobe (Figure 1). Brain magnetic resonance imaging (MRI) at 20 months of age (Figure 2) showed focal gyral atrophy in the right anterior frontal lobe on T1 and T2 weighted-images associated with minimal gliosis on FLAIR sequence. Gradient Echo demonstrated blooming artifact corresponding to the calcification seen on brain CT. In addition, prominent focal leptomeningeal enhancement was seen on post contrast T1 weighted-images.

We started him prophylacticaly on leveteracitam and aspirin. Follow up six months later did not reveal any new recurrences of seizures.

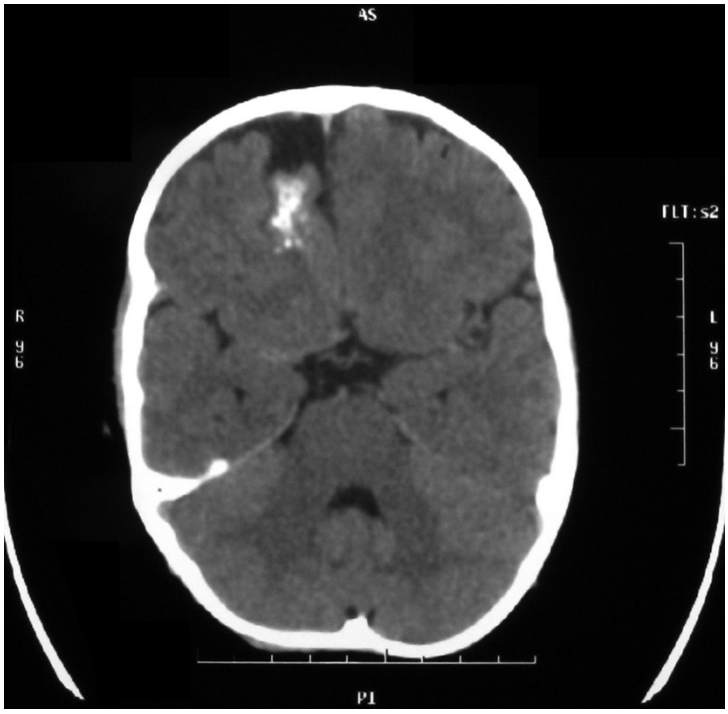


**Figure 1: Axial non- enhanced CT scan of the brain showing focal gyral atrophy at the right anterior frontal lobe associated with prominent subcortical calcification**

**Case 2**

A six-year old female child presented with a febrile focal left sided seizure at the age of eight months was examined. Since that time, she continued to experience frequent febrile and afebrile focal and generalized seizures. She had school difficulties; however, her general ophthalmology and neurological exam were normal.

Initial neuroimaging at the age of one year which included non-enhanced brain CT scan (Figure 3 A) revealed focal cortical atrophy at the right superior fronto-parietal lobe without calcifications. Follow up brain MRI at the age

of two and half years old showed focal atrophy of the right fronto-parietal region (Figure 3B).

Her last seizure at the age of six years was associated with focal post ictal weakness that lasted more than 24 hours, which necessitated the repetition of her neuroimaging. Her last brain MRI (Figure 3 C) at six years of age revealed focal gyral atrophy in the frontoparietal region with prominent leptomeningeal enhancement. Brain CT did not show calcification.

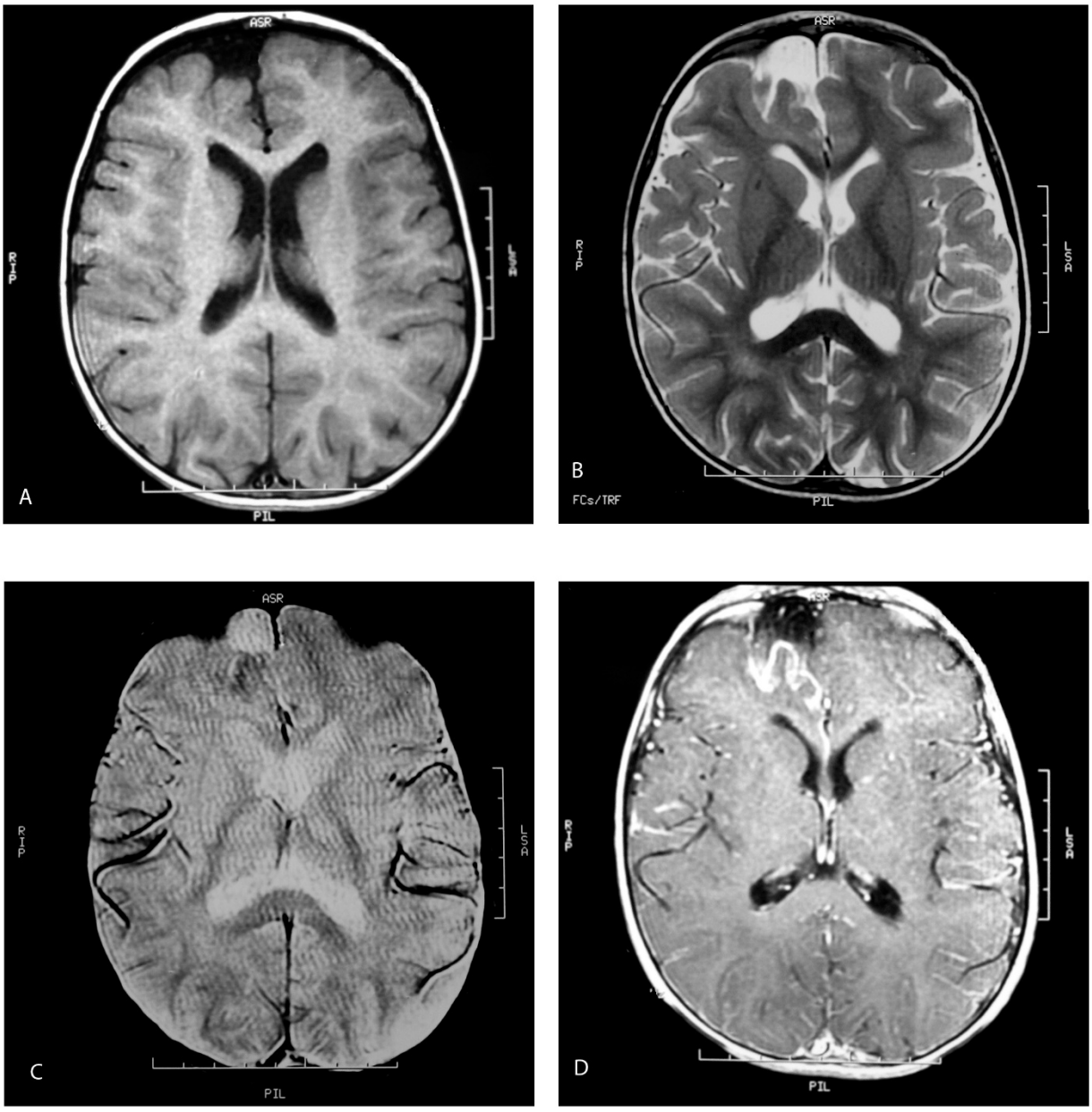


**Figure 2: Axial T1 (a) and T2 (b) Weighted-images showing focal gyral atrophy at the right anterior frontal lobe. Axial Gradient Echo image (c) demonstrating subcortical blooming artefact reflecting calcification seen on CT. Prominent focal leptomeningeal enhancement over the right anterior frontal lobe is seen on axial T1W post contrast image (d)**

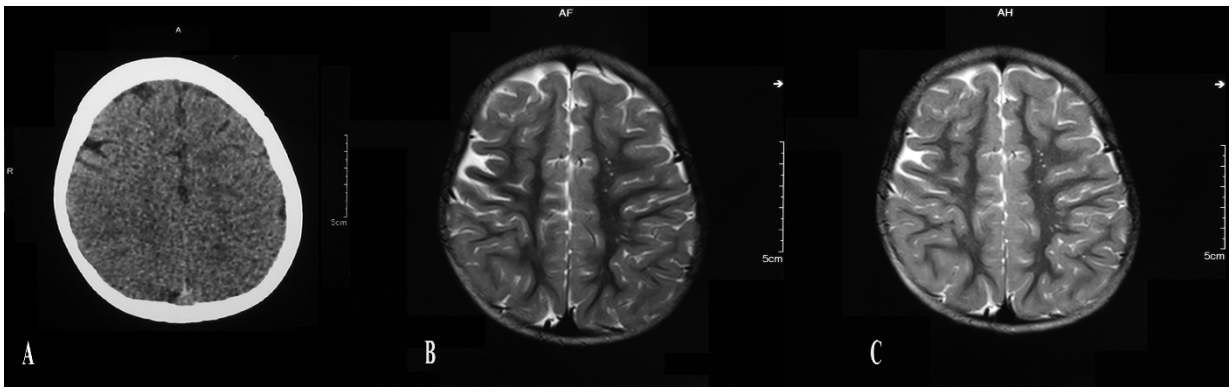


**Figure 3: (A) Initial axial non-enhanced brain CT scan showing focal cortical atrophy at the right superior fronto-parietal lobe without calcifications**
**(B) 1.5 years later follow up Axial T2W showing focal cortical atrophy at the right superior fronto-parietal lobe**
**(C) Last MRI T1W with contrast demonstrating additional prominent leptomeningeal enhancement**

**Discussion**

SWS without facial nevus is a rare disorder which needs a high index of suspicion. Our two patients had seizures provoked by fever. While seizure might seem at the beginning to follow a mild course, aggressive prophylactic treatment is needed because of the progressive nature of the disease[9]. Although patients without facial angiomas are known to have milder impairment of vascular perfusion,the cortex will not be able to handle the extra metabolic burden with the onset of recurrent seizures[9]. The leptomeningeal angiomas might even be absent on initial neuroimaging and may appear only on later imaging due to the increased metabolic demand from seizures[9].

Magnetic resonance imaging, specifically T1 weighted imaging with gadolinium contrast, together with susceptibility-weighted imaging (SWI) is the recommended imaging modality for demonstrating characteristic findings of SWS[10].

Our first patient showed typical atrophy, subcortical calcifications, and leptomeningeal enhancement of limited extent and isolated to the right frontal lobe anteriorly. The frontal lobe location of the leptomeningeal angioma with focal isolated involvement of its anterior portion in our patient is very unique. To the best of our knowledge, there are only three cases in the English literature of patients with SWS without facial nevus and with frontal lobe involvement[4,7,11]. In all three cases, the angiomas extended to involve the parietal and/or temporal lobes, while in our first patient it is confined only to the frontal lobe. Nevertheless, follow up imaging in our patient might be necessary as progression might appear on future neuroimaging. In addition, our second patient had a rare later appearance of a frontal leptomeningeal angioma that adds more to the existing few literature on frontal leptomeningeal angioma in SWS[4,7,11]. Reporting more patients might help delineate the clinical picture and prognosis in these patients.

In conclusion, we reported two cases of SWS with rare location of leptomeningeal angiomas.

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# متلازمة ستيرجي ويبر بغياب وحمة الوجه: وصف لحالتين

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**الملخص**

تعتبر متلازمة ستيرجي ويبر من الامراض العصبية النادرة وتتميز بوجود ورم وعائي بالسحايا الرقيقة بالإضافة لوجود وحمة حمراء على الوجه وقد يصاحب هذه المتلازمة أيضا ارتفاع بضغط العين.

تهدف هذه الدراسة لوصف سريري وشعاعي لمريضين مصابين بمتلازمة ستيرجي ويبر بغياب وحمة الوجه الحمراء حيث يعتبر غياب الوحمة نادراً جداً كما وتناقش هذه الدراسة الحالات المشابهة التي وردت في الابحاث السابقة.

**الكلمات الدالة:** متلازمة ستيرجي ويبر، وحمة الوجه، ورم وعائي متعلق بالسحايا الرقيقة، الفص الجبهي.