CASE REPORTS

STURGE-WEBER SYNDROME (SWS) WITH OCCIPITAL EPILEPSY: TWO CASES PRESENTED IN A TERTIARY CARE HOSPITAL

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Abstract

Sturge-Weber syndrome is a rare sporadic, neurocutaneous syndrome. In most instances, the patients present with seizures other than the port-wine stain and ipsilateral leptomeningeal angiomata. The documentation of seizures' semiology in the current literature is scarce. Here we reported two cases who presented with visual hallucination as an ictal phenomenon and associated ictal headache and vomiting. The phenomenology was very close to migraine with aura. A careful history, associated seizure, and EEG findings helped in differentiation. Both the cases responded well with carbamazepine.

Keywords: *Sturge-Weber syndrome, occipital epilepsy, migraine with aura.*

DOI: *https://doi.org/10.3329/jdmc.v30i1.56912*
J Dhaka Med Coll. 2021; 30(1) : 118-222

Introduction:

Sturge-Weber syndrome is one of the disorders of Phakomatoses.[1] The presence of a triad in its complete form characterizes the disease.[2] The triad includes the presence of a facial port-wine stain (facial capillary malformation) mainly involving the ophthalmic division of the trigeminal nerve, ipsilateral leptomeningeal angiomata (most often involving the occipital and posterior parietal lobes), and angioma involving the ipsilateral eye.[3] This disease is sporadic, and the GNAQ gene on chromosome no 9 causes the disease.[4] This mutation influences the RAS effector pathways thereby increases the proliferation and inhibits apoptosis.[2] There are three types of Sturge-Weber syndrome.[5] In classical form or type one, all the triad components are present. In type two, there is an absence of intracranial leptomeningeal angiomata, and in type three, there is the presence of only intracranial leptomeningeal angiomata.[5] The presentation of the disease varies in terms of age of onset and the associated presenting features.[6] The cardinal manifestations includes seizures (75% to 90%), progressive hemiparesis (25% to 60%), headache mimicking migraine (30% to 45%), cognitive development (50% to 60%) [2, 7]. The documentation of the pattern of seizures or ictal semiology is scarce in the current literature. Occipital epilepsy manifests with occipital seizure, which may be idiopathic, cryptogenic, and symptomatic. The Sturge- Weber syndrome is one of the rare symptomatic causes of occipital epilepsy [8]. Here we reported two cases of Sturge-Weber syndrome who presented to the headache clinic of Dhaka Medical College with occipital ictal semiology. We obtained informed written consent from the patient in the first instance and the patient's mother in the second case.

Description of the cases:

Case 1:

A lady of 60 years with port-wine stain since her childhood came to epilepsy clinic, Dhaka Medical College, with a headache for two years. The headache was preceded by visual hallucinations 15-30 minutes before the onset of the headache. She described the hallucination as multicolored lights with occasional flushing of lights. The headache was throbbing in nature, severe in intensity, and persisted for 12-24 hours. The visual

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Received: *01-01-2021* ***Revision:*** *03-02-2021* ***Accepted:*** *01-03-2021*

hallucination continued till the headache ended, and after that, she complained of transient blurring of the vision for about 15-20 minutes. She had associated severe vomiting. There was associated hemiparesis at the peak of the headache, which persisted for 30 minutes. The visual hallucination and headache occurred 2-3 times per month. She had a single episode of generalized seizure three months back. Her milestones of development were within normal. There was no family history of such type of illness. She had an average intellect. Her port-wine stain was pink in color, well-demarcated, confined in the forehead with associated hypertrophy of the overlying skin. Fundoscopic examination revealed choroidal hemangioma in the left nasal quadrant. There was no glaucomatous change, and her intraocular pressure was normal. CT scan of the head revealed calcification in the left parieto-occipital region. EEG showed the absence of alpha in the left hemisphere and focal epileptiform

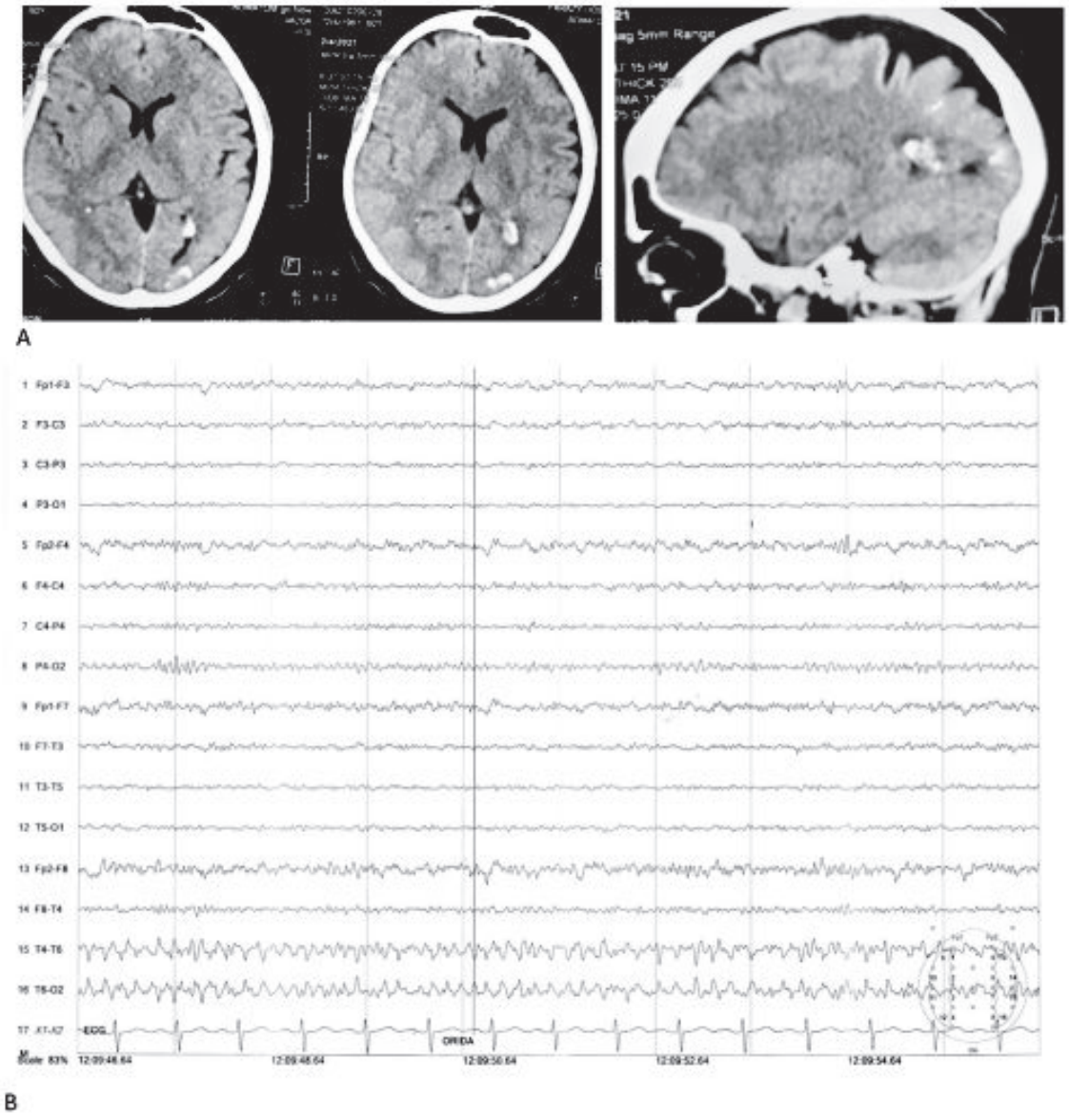

Figure 1 : *A. Non contrast CT of head with gyriform subcortical calcificaiton in left parieto-occipital region with focal cortical artrphy*

B. EEG with asymmetric background (loss of alpha in left hemisphere) and in right side superimposed spike with phase reversal on posterior bipolar montage.

discharges from the right hemisphere. She was given tab carbamazepine 200 mg twice daily, and after one month of follow up the headache intensity and the hallucination was diminished to 1 attack per month. The dose of carbamazepine was escalated to 300 mg daily. At four months of follow-up, she was free from headaches and the visual phenomenon as well.

Case 2:

A 2-year-old child visited the epilepsy clinic of Dhaka Medical College with a severe headache for six months. The headache occurred 2-3 times per week. A frightening hallucination and photophobia, which persisted for a few seconds, happened before the headache. After that, the headache started with vomiting, and the baby started crying for 2-3 hours. She also had several episodes of generalized seizure. The milestones of her development were within normal. There was a port-wine stain over the left face in the maxillary and ophthalmic division of the trigeminal nerve, with no overlying skin hypertrophy. Her ophthalmologic examination was not possible. CT head revealed gyriform calcification and Hemi atrophy of the right parieto-occipital region. She was given syrup carbamazepine 10 mg per kg/day in three divided doses. At the one month of her follow-up, the headache intensity and the severity were decreased to 50%. We escalated the drug dose to 15 mg/kg/day and advised her to follow up after one month.

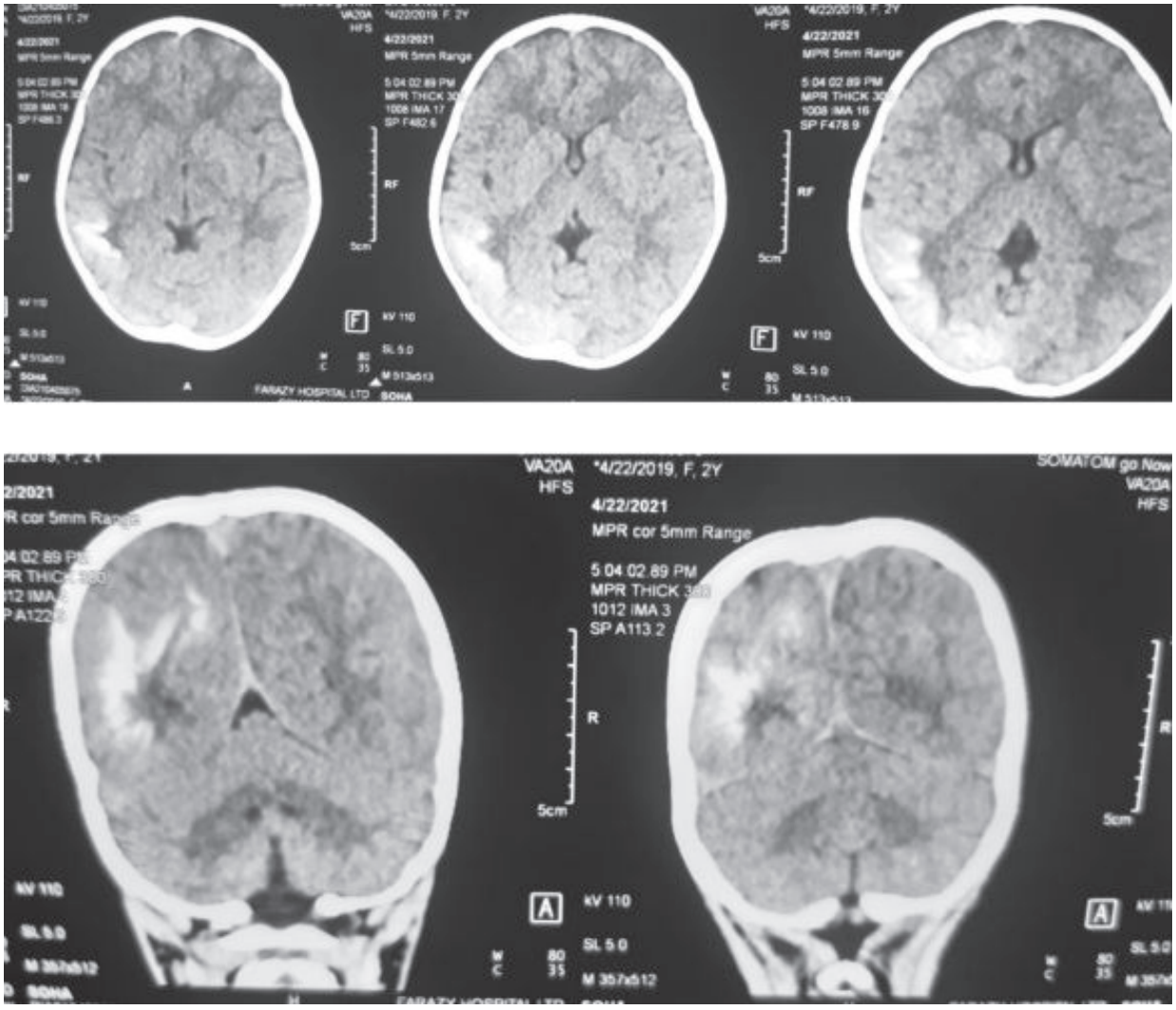

Fig.-2: *Non-contrast CT of Head: subcortical gyriform calcification in the right parieto-occipital region*

Discussion:

Both cases came to us with visual hallucination, which can be explained as occipital seizure semiology. They also had ictal headache and vomiting with an occasional secondary generalization of the seizure.

None of the cases had a family history of Sturge-Weber syndrome, which indicates its sporadic nature.[4] The port-wine stain in both instances presented from birth. In the first instance, it was confined to ophthalmic division only, but in the second instance, it involved both maxillary and ophthalmic division of the trigeminal nerve. In both cases, the color of the port-wine stain was purple, and the margin was well defined, so we could easily differentiate it from salmon patches (nevus simplex).[9] In both patients, glaucoma was absent, which might present in 8-15% of the instances.[10] In the presence of the leptomeningeal angiomata, there is a 90% Chace of the development of seizure.[2] The associated seizures are usually focal or focal with impaired and focal with bilateral generalization.[11] In about 75% of the instances, the convulsions start in the first year. In 90 % of cases, it begins within two years of life.[12] In our second case, the seizure had started at two years, but in the first case, the convulsion occurred in the fifth decade.

Both the cases had a headache and visual hallucination mimicking the visual aura of migraine. We can differentiate the ictal semiology of occipital epilepsy from migraine by its character, duration, and associated symptoms due to progression.[13] Frightened brief visual hallucination with generalized seizure in the second case and a multi-color elemental simultaneous visual hallucination with headache, luminance within a visual territory, and transient ictal blindness in the first case were the differentiating features from the aura of migraine. We can explain the headache and vomiting by ictal headache and vomiting, which might present in 50% of instances.[13] In migraine with the aura, the aura persists for 5-60 minutes, occurs before the headache onset, and subsides after the headache onset. The migraine aura is mainly uncolored and linear.[14]

The calcification can be detected at even one year of age. It is mainly due to dystrophic calcification resulting from hypoxia.[2] In both instances, there was calcification in the typical occipital and posterior parietal region [fig-1 A, fig-2]. The seizure might be due to cortical irritation resulting from hypoxia, ischemia, or gliosis.[11] We could not perform EEG and fundoscopy in the second case. But in the first case, the EEG with interictal unilateral loss of background alpha and unilateral spike were consistent with occipital seizure [8]. In both of the instances, the patient responded well with carbamazepine.

Conclusion:

The migraine-like headache in Sturge- Weber syndrome should be evaluated carefully for occipital epilepsy. It will help in the proper selection of the drug for these patients.

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