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Case Report

Sturge-weber syndrome: a case report with complex presentations

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ABSTRACT

Sturge-Weber syndrome (SWS), a rare, congenital phakomatosis is the fourth among the neurocutaneous syndromes and is characterized by facial nevus flammeus, leptomeningeal angioma and glaucoma. It is also manifested as cutaneous, neurological, ophthalmic and behavioral symptoms. It is diagnosed primarily via cerebral abnormalities noted in MRI and CT scans. This case report discusses the complexity of Sturge-weber syndrome with conditions such as Hepatitis A and sickle cell anemia. We report a case of a 11-year-old male patient presented with Convulsions and birthmark over left-side of the face. SWS was confirmed by left cerebral atrophy while HPLC and Hepatitis A virus IgM antibody suggested sickle cell trait and Hepatitis A respectively. Symptomatic treatment was given and the patient was counseled to adhere to the anti-epileptics. Additionally, dye-laser photocoagulation was suggested as an intervention to remove port-wine facial birthmark. SWS is a rare phakomatosis and diagnosis can be done via characteristic symptoms and Imaging techniques. As it is incurable, emphasis is placed on proper prognosis and symptomatic treatment, with a focus on managing co-morbidities.

Keywords: Neurocutaneous syndromes, Seizures, Hepatitis, Case report, Sickle cell, Port wine stain

INTRODUCTION

Sturge-weber-syndrome (SWS) is one of the phakomatoses affecting primarily the microvasculature of face, eyes and the leptomeninges. It is the fourth among the other phakomatoses.[1] SWS was designated after William Sturge and Frederick Weber who explained in description, the features of this disease.[2] It is a rare disorder with estimated prevalence of 1: 50000. Nevus flammeus is the primary manifestation progressing to seizures and glaucoma. It may also cause headache, stroke-like symptoms and developmental delays.[3]

The Roach scale classifies SWS into 3 categories namely; type-1 with angiomas involving the face and leptomeninges, angiomas may develop glaucoma, type-2 with only facial angioma, may develop glaucoma and type-3 is leptomeningeal angioma alone with absence of glaucoma.[4] It is caused by non-germline mutation of the Guanine nucleotide-binding protein G (q) subunit alpha gene (GNAQ) sporadically in cells of vascular plexus causing excess production of cells in cephalic microvasculature resulting in angiomas.[5] It is diagnosed via cerebral atrophy with widened subarachnoid space seen in magnetic resonance imaging (MRI).[6] The condition is treated symptomatically with carbamazepine as drug of choice in seizures and ophthalmic beta-blockers for glaucoma. Since the syndrome is untreatable, the well-being of the population with this syndrome should be improved by symptomatic treatment. Here, we provide a case report concerning an 11-year-old male diagnosed of SWS.

CASE REPORT

A 11-year-old male patient presented with complaints of convulsion since 1 day, fever, cough and abdominal pain since, 2 days and icterus. During convulsion, left side

upper and lower limb stiffness with up rolling of eyeball was noted. Patient was on oxcarbazepine 150 mg and clobazam 5 mg. The first febrile seizure occurred when patient was 1 year old whereas second episode occurred after 3 months and was diagnosed as right focal epilepsy. Since birth, the patient presented with a prominent Hemangioma over left side of the face which extended from the forehead down to the cheek reaching towards the jawline (Figure 1). MRI Brain showed mild left cerebral atrophy with enhancement of parieto-occipital region and enlarged choroid plexus suggestive of Sturge weber syndrome. The patient had positive sickling with HPLC suggesting SCT.

Laboratory tests showed anemia (hemoglobin 11 g/dl), hyperammonemia (275 µg/dl), high C-reactive protein (56.6 mg/l) (CRP), high liver function test (LFT), hypoalbuminemia (serum albumin 2.7g/dl) and high anti HAV IGM (18.56 index) (Table 1). Ultrasonography (USG) showed mild hepato-splenomegaly and minimal ascites. Laboratory values are elaborated in the table (Table 1).

On admission, levetiracetam PO (3.5 ml/50 ml normal saline (NS) Stat) was given to control seizures, paracetamol IV (2.3 ml/5 ml NS SOS), ceftriaxone IV (10 ml/10 ml NS twice daily), oxcarbazepine PO (150 mg twice daily), clobazam PO (5 mg half tablet once daily), pantoprazole IV (40 ml/5 ml NS once daily), ondansetron IV (1.7 ml/ 2 ml NS SOS), tobramycin and carboxymethylcellulose eyedrops were prescribed. On 5th day, metronidazole IV (15 mg/kg thrice daily), ursodeoxycholic acid 300 mg, rifaximin 200 mg, multivitamin and folic acid were given PO once daily and vitamin D3 sachet was given once a week. Lactulose syrup 15 ml thrice daily was also prescribed as patient had constipation.

Vitamin A (4 tablets of 50,000 U once daily), vitamin E 1 capsule, vitamin K IV (5 mg) were given from 5th day. The patient had an episode of vomiting on 7th day and fever spike on 2nd day. The patient got symptomatically better with no recurrent seizures and was discharged with levetiracetam (120 mg), oxcarbazepine (150 mg) folic acid (5 mg) to be continued orally once daily while clobazam (5 mg) were given orally twice daily.

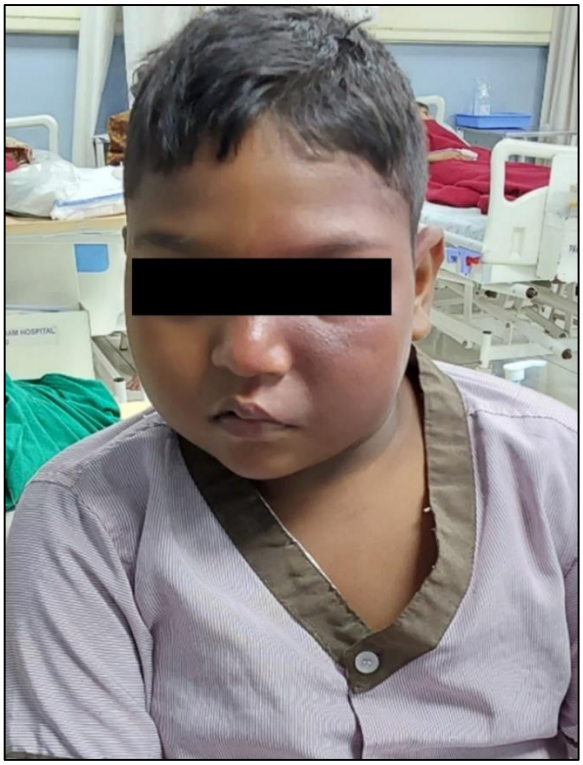

Figure 1: Port-wine stain over left side of the face.

Figure 1 shows the 11-year-old patient with a port-wine stain extending from the forehead to the cheek and ending at the jawline affecting a substantial portion of skin. This port-wine stain is suggestive of Sturge-weber syndrome.

Table 1: Abnormal laboratory parameters of the patient.

Parameter	Observed value	Normal value
Hemoglobin	11 (↓)	13–17 g/dl
Hematocrit	32.2 (↓)	36–40%
Neutrophil	61 (↑)	32–54 %
Ammonia	275 (↑)	30-150 µg/dl
CRP	56.6 (↑)	< 6 mg/l
ALT	2164 to 1445 (↑)	Up to 45 U/l
AST	7090 to 1157 (↑)	Up to 35 U/l
Total bilirubin	6.1 (↑)	0-2.0 mg/dl
Direct bilirubin	3.8 (↑)	0.0-0.4 mg/dl
Indirect bilirubin	2.3 (↑)	0.0-0.6 mg/dl
Alkaline phosphatase	315 (↑)	53-128 U/l
Total protein	5.80 (↓)	6 - 8.3 g/dl
Serum albumin	2.7 (↓)	3.5 - 5.2 g/dl
HAV IgM	18.56 (↑)	>1.1 index
Fundus examination	No signs of Papilledema	

DISCUSSION

SWS, also known as Sturge weber Dimitri syndrome is one of the major neurocutaneous syndromes causing angiomas over face, eyes and leptomeninges. It equally affects male and females. It is classified via the Roach scale. SWS occurs due to mutation in GNAQ gene on the chromosome 9 causing hyperplasia of the cells in the microvasculature, resulting in angiomas. The typical manifestation is unilateral nevus flammeus extending bilaterally with seizures. The diagnosis is usually done via symptoms and confirmed with radiological investigations. MRI with gadolinium contrast and computed tomography show cerebral shrinkage while X-Rays show tram-track sign.

Fundus examination is done to check signs of papilledema. Treatment for SWS is mainly symptomatic, focusing on managing seizures with medications like Carbamazepine, glaucoma with beta-blocker eye drops, and other supportive therapies for associated symptoms like headaches and stroke-like episodes. Surgical intervention such as lobectomy, vagal nerve stimulation, corpus callosotomy, focal resective surgery could be recommended for patients with refractory seizures, trabeculectomy for glaucoma while dye laser photocoagulation can be advised to remove port-wine stain (PWS).[7] Modified Atkins diet may also be suggested to the patients with refractory epilepsy. Prognosis is dependent on early identification of symptoms.[8]

Hepatitis A is the liver inflammation caused by HAV whereas SCT is the inheritance of HbS from one parent. While there is no advanced research on the association of these conditions, reperfusion therapy in SCT can make an individual prone to viral infections.[9,10] While there is no direct correlation between hepatitis A, SCT and SWS, dysregulated immune responses due to SCT may potentially influence the progression or severity of SWS.[11] The management of such complex cases involves tailored treatment strategies for each condition, focusing on controlling seizures, addressing liver inflammation, and managing the hematological aspects of SCT.[12]

The patient in our report is Type I of Roach scale due to the presence of seizures and PWS. Zallman et al, emphasized the characteristic features such as cutaneous capillary malformations (PWS) and cerebral capillary malformations, which align with the clinical presentation in the reported case.[13] The presence of mild left cerebral atrophy in MRI with enlarged choroid plexus confirmed SWS. Furthermore, the patient was diagnosed with SCT and Hepatitis A confirmed with laboratory findings of anemia, HPLC, hyperammonemia, high CRP, high LFT, hypoalbuminemia, high Anti HAV IgM and Hepato-splenomegaly in USG, leading to a complex clinical scenario requiring a multidisciplinary approach to management.[14] The case report by Lopez et al, on subdural hematoma in a patient with SWS sheds light on potential complications associated with the syndrome thus, highlighting the importance of vigilance in monitoring for such complications, especially in cases with additional health conditions like Hepatitis A and SCT.[15]

The patient was treated for seizures with levetiracetam while ursodeoxycholic acid and rifaximin were given for hepatitis A and other treatment plan for symptomatic relief. On discharge, the patient was advised to be compliant to the Anti-epileptic medicines and Dye-laser photocoagulation was suggested to remove port-wine facial birthmark as the patient was little bit ashamed being a teenager with a big facial birthmark. On follow-up, the seizures were in control with the resolution of Hepatitis A. These management strategies emphasize the need for tailored treatment plans based on the Roach scale classification of SWS, similar to the approach taken in the case report by Karim et al.[16]

CONCLUSION

SWS is a rare neurocutaneous syndrome causing facial, ophthalmic and leptomeningeal angiomas. SWS pose challenges due to its multisystem involvement and lack of curative treatment, necessitating a comprehensive and individualized approach to patient care. The presented case underscores the importance of a holistic care plan for individuals with rare conditions like SWS, especially when complicated by additional comorbidities. Early identification with proper prognosis is essential for the diagnosis of SWS to avoid complications.

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