International Clinical and Medical Case Reports Journal
Case Report (ISSN: 2832-5788)

Case Report of Sturge Weber Syndrome With Bilateral Port Wine Stain And Bilateral Cerebral Calcification

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Citation: *Tharuna Chandra, Govind Pandey, Gaurav Singh. Case Report of Sturge Weber Syndrome With Bilateral Port Wine Stain And Bilateral Cerebral Calcification. Int Clinc Med Case Rep Jour. 2024;3(6):1-5.*

Received Date: 24 June, 2024**; Accepted Date:** 26 June, 2024**; Published Date:** 28 June, 2024

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ABSTRACT

Sturge-Weber Syndrome (SWS) is a congenital disorder affecting the brain, eyes, and skin. It is characterized by a facial cutaneous vascular nevus, angioma, and bupthalmos. The frequency of SWS is between 1:20,000 and 1:50,000. There are very few reported cases of SWS with bilateral intracranial calcification and bilateral port wine stain. We present a case report of an 8-year-old boy present with complaint of generalized tonic-clonic (GTCS) type of convulsion with purple discoloration of the skin on the both side of the face since birth and was diagnosed with SWS with bilateral intracranial calcification, intellectual disability, seizure and bilateral port wine stain. MRI with contrast revealed gyriform hyperdense calcification is seen in bilateral fronto-parietal lobes and T2 suggestive of calcification seen in bilateral fronto- parietal lobes with prominence of supratentorial sulcal- cisternal spaces seen on right side. The patient was counseled about the syndrome and discharged on anti-convulsant treatment with advice for dye laser photocoagulation for port-wine stain.

Keywords: Neurocutaneous syndrome; Port-wine stain; Sturge weber syndrome (SWS); Nevus flammeus; Tram track appearance

INTRODUCTION

Sturge–Weber Syndrome (SWS) is a congenital, non-hereditary disorder affecting the brain, eyes (commonly) and skin. It is also known as encephalotrigeminal angiomatosis characterized by a facial cutaneous vascular nevus (nevus flammeus or port wine stain) in association with facial angioma and bupthalmos. It is a neurocutaneous syndrome characterized by angioma in the leptomeninges (pial) and facial skin port wine stain, usually in the distribution of ophthalmic and maxillary parts of trigeminal nerve.[1] 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. The frequency of occurrence of SWS is between 1:20,000 and 1:50,000.[2] We present a case report of an 8-year-old boy diagnosed with SWS with bilateral intracranial calcification, bilateral port wine stain, intellectual disability and seizure.

CASE ILLUSTRATION

A 8- year old male being managed as case of epilepsy reported to the Department of Pediatrics, BRD Medical College, Gorakhpur with the chief complaints of fever for 5 days and following day witnessed two episodes of tonic-clonic seizure with secondary generalization followed by altered sensorium. There was no associated aura, headache, vomiting, head trauma, stroke like events or diminished vision and no history of meningism. He was full term born following uneventful birth events. His past medical history revealed he was a known case of epilepsy and on irregular long term antiepileptic treatment with oral phenobarbitone since 1 year of age. He had a similar episode at the age of 3 years and was started on valproate. His family history was non contributory.

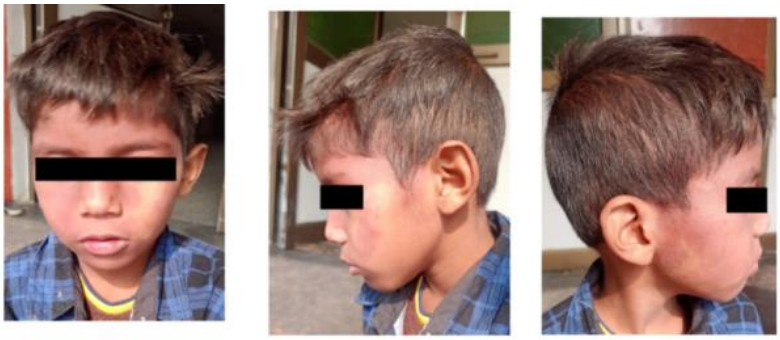

Figure 1: Port-wine stain (front, left & right views) extensively distributed bilaterally on forehead, left & right cheek, jaw and lips.

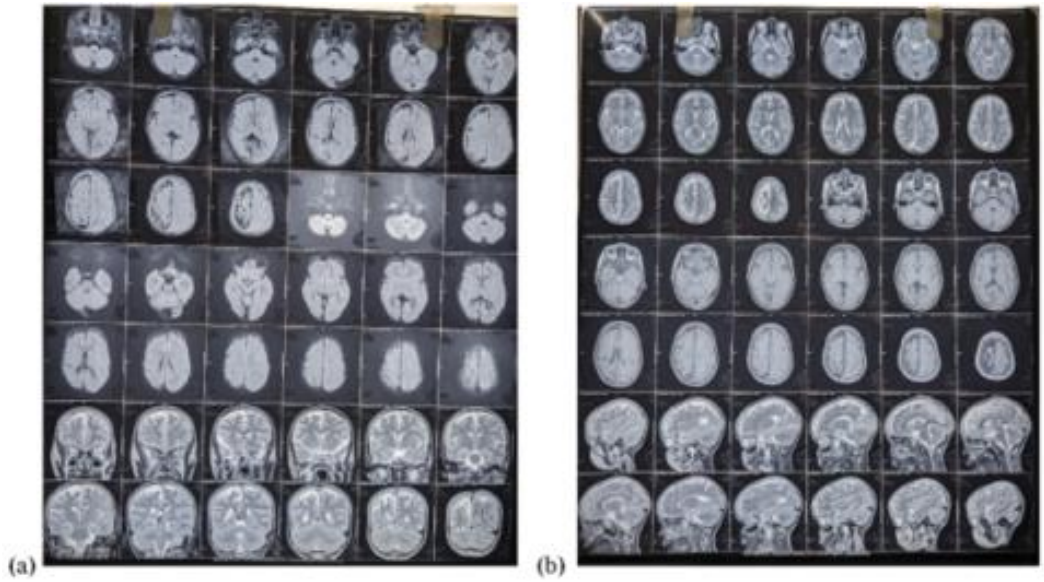

Figure 2 (a) & (b): Magnetic resonance imaging of the brain reveal gyriform T2 hypointense foci of blooming s/o calcification are seen in bilateral fronto- parietal lobes with prominence of supratentorial sulcal- cisternal spaces seen on right side.

Examination findings on presentation revealed a male school-going child with a moderate intelligence quotient without any evidence of cardiovascular and respiratory disease. He had a bilateral purplish red patch centered around his forehead, eyes, side of the nose, and upper lip extending to the midline in the distribution of

ophthalmic and maxillary division of trigeminal nerve, suggestive of port-wine stain. He was unconscious with a Glasgow Coma Score of 10/15. He had no weakness in limbs and bilateral extensor plantars. There was hypertonia and hyperreflexia. Based on the present finding, a provisional diagnosis of Sturge-Weber syndrome was made. He was commenced on intranasal oxygen, antibiotics, antipyretics, and AED (loading and maintenance). He became seizure-free and gradually regained consciousness. The patient was advised to go for an ophthalmic examination, which revealed mild nasal blurring of disc margins in both eyes. Blood examinations were normal. Brain CT scan revealed diffuse hyperdensity in both cerebral hemispheres with calcification and Tram track appearance. Electroencephalography was suggestive of a borderline abnormal sleep pattern in which intermittent slow-wave discharge was present. MRI with contrast revealed gyriform hyperdense calcification in bilateral fronto-parietal lobes with prominence of supratentorial sulcal-cisternal spaces seen on the right side. Blooming on the SW1 sequence is seen in the right parasagittal occipital region, suggesting leptomeningeal angiomas. The radiological findings confirmed the diagnosis of SWS. Psychological examination revealed an IQ of below 70 with aggressive tendencies and emotional imbalance.

Neurologic examination, including cranial nerves, motor, sensory, and cerebellar examination, was within normal limits and revealed no neurologic deficits. Patient attendants were counselled about the syndrome and patient was discharged home on two anti convulsants treatment with advice for dye laser photocoagulation for the port wine stain and nutritional supplements after two weeks of admission.

DISCUSSION

SWS is a rare congenital neurocutaneous disorder. Etiology is not well established, but a study has shown that a somatic activating mutation in the GNAQ gene is responsible for SWS.[3] Schirmer described the first case, and then in 1879, William Allen Sturge gave a more precise description of the disease to neurologic symptoms.[4] Frederick Parkes Weber, in 1929, elucidated the radiologic features seen in patients of SWS.[5] When the upper and lower eyelids are involved in port-wine birthmark, the risk of developing glaucoma becomes as high as 50%.[6] In 1992, Roach categorized SWS variants into three types:

1. Type I: The individual has a facial PWS, leptomeningeal angioma, and may have glaucoma
2. Type II: The individual has a facial PWS, no leptomeningeal angioma, and may have glaucoma
3. Type III: The individual has leptomeningeal angiomatosis, no facial PWS, and, rarely, glaucoma.[7]

Our case is consistent with Type I of SWS variant. Besides, leptomeningeal hemangiomas can be present, and they cause the atrophy of the cortical parenchyma of the brain, seizures, migraine, and cognitive impairment.[8] The exact reason for the vascular and neurologic involvements remains an ongoing debate; however, it is hypothesized that capillary malformations (CM) and leptomeningeal angiomatosis could result from the absent regression of a primitive cephalic plexus.[9] The persistence of primordial sinusoidal vascular channels and the underdevelopment of the brain's superficial venous drainage with compensatory deep dilated venous channels also affect the capillaries. A close association between neurological affection and the severity of cutaneous involvement in SWS has been described in the past. However, more recent studies have shown that neither the size nor the distribution of facial PWS correlates with neurological features. Seventy-five to 90% of children with SWS develop partial seizures by three years of age, and none demonstrate that early onset of seizures indicates a poor prognosis. However, some patients develop intractable epilepsy, hemiatrophy, visual field cuts,

and mental retardation (50-75%).[10] In our case, the seizure attack was from the age of 1 year, and the port wine stain was from birth itself. As the time went by child showed mental retardation and emotional lability. Fever and infection often precipitate a seizure onset, which is consistent with our patient history. Sujansky et al. suggest that mental retardation may be more common in children whose seizures begin before the age of 2 years or who have seizures that are not controlled with antiepileptic drugs.[11] The extent of neurologic involvement dictates the degree of developmental delay and intellectual disability in SWS patients. Delays are much more common in patients with bilateral disease. Approximately 50-60% of patients with SWS will have developmental delay or mental retardation, or both. Anecdotally, patients who develop debilitating aspects of SWS often do so before grade school; hemiparesis, hemianopia, retardation, and epilepsy are often apparent but infrequently severely worsen before grade school. Attention-deficit hyperactivity disorder is another comorbid condition associated with SWS. As with Attention deficit hyperactivity disorder (ADHD) patients in the general population, SWS patients need to be monitored for medication requirements to minimize impulsivity and inattention. Management of attention-deficit hyperactivity disorder can be achieved and may significantly improve daily functioning in some SWS patients. Lifelong medical treatment coupled with frequent surgeries is the standard. Early surgical treatment controlled the seizures, but other neurological problems, such as hemiparesis and intellectual deficits, showed a less satisfactory response. Making a diagnosis requires clinical evaluation, and it is confirmed by imaging modalities such as plain X-ray, Computed Tomography (CT), and Magnetic Resonance Imaging (MRI) scan. Treatment is mainly symptomatic and supportive, with control of seizures using antiepileptic drugs. Surgical resection is reserved for intractable cases of seizures. Although there is no conclusive evidence that surgical management in infancy provides a better prognosis, delay of surgical treatment may result in further cognitive deterioration.[12] Our patient, though, was counseled for the need for epilepsy surgery like hemispherectomy, and for the neurobehaviour, methylphenidate, and clonidine may be tried on further evaluation of the patient. Early onset of seizures and poor response to medical treatment, bilateral cerebral involvement, and unilateral severe lesions were indicative of a poor prognosis. The epileptic phenomena of SWS can manifest with atypical psychiatric symptoms and need correct identification and appropriate response for optimal management. Thus, appropriate psychiatric counseling of patients and attendants is essential.

CONSENT: Written informed consent was obtained from the patient for publication of this case report.

ACKNOWLEDGEMENT: None

CONFLICT OF INTEREST: None

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