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Case Report

Sturge Weber Syndrome Roach Type I: A case report

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Abstract

Convulsion is a serious neurological symptom with diverse underlying etiology ranging from metabolic causes to organic brain lesions. Congenital neuro-cutaneous syndromes are rare causes of seizures that manifest with specific external features. This article reports a 22-year-old man born to non-consanguine parents, who presents with recurrent seizures commencing at the age of one year along with thephysical sign of capillary hemangioma on the right side of theface and radiological evidence of ipsilateral subcortical calcification in the brain. These features constitute the diagnosis of a rare neuro-cutaneous disorder, the Sturge-Weber syndrome, a classical variant, the Roach Type I.

Key words: Sturge-Weber Syndrome, neuro-cutaneous syndrome, port-wine stain,leptomeningeal angiomas, Roach's scale

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Introduction

Sturge-Weber Syndrome (SWS), a rare angiomatosis, is characterized by congenital angiomas of lepto-meningesand the face.[1]Though SWS is the most popular name for the condition, it is also known as Phakomatoses or 'mother-spot disease' and encephalo-trigeminal angiomatosis.[2]This is a rare condition but the third in order among all neuro-cutaneous syndromes, after tuberous sclerosis and neurofibromatosis.[3] Schirmer was the first to identify and describe this condition in 1860, but William Allen Sturge pointed out in 1879 that, the triad of involvement is in the face, central nervous system, and eyes in its complete manifestation, and its radiological characteristics were elaborated by Frederick Parkes Weber. The name SWS goes after their names.[1, 2] Its overall prevalence is 1 in every 20-50,000 live births and from an embryologic point of view, it results from non-regression of the cephalic part of the primitive venous plexus around the fetal neural tube.[4,5] As depicted by Shirly et al, SWS occurs due to somatic activating mutation in the GNAQ on the 9th chromosome. However, it develops sporadically and lacks a specific inheritance pattern. This gene plays a role in synthesizing the protein G alpha subunit q, which controls cell proliferation and enhances apoptosis.[1,5] The genetic mutation shows a mosaic pattern of affection and is found to be prevalent in the tissues involved. The genetic basis of SWS has been confirmed by many other researchers and some of them have shown that eighty percent of the affected individuals have a specific genetic mutation in the brain.[6] Clinical manifestations can be variable depending upon the organ involvement. Facial capillary hemangioma called port-wine stain (PWS), found along the distribution of the 5th cranial nerve, ocular angiomas causing glaucoma, and, leptomeningeal angiomas causingseizures, constitute a complete form of SWS. It can be of three clinical types according to the Roach scale-types I, II, and IIIbased on the presence of angiomas of the face, brain, and/or eyes.[7]

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Case Report

A 22-year-old Muslim, unmarried male from Moulvibazar, working in a private company, was brought to the Medicine inpatient of Sylhet Women's Medical College Hospital, with convulsion for 1 day. According to his sister, who had been a witness to the event, it started from his left leg and then rapidly involved the left side of his body. It persisted for several minutes and then faded away for several hours and came back again. The convulsion kept occurring repeatedly for one day until he got admitted to the hospital. However, there was no history of fever, neck stiffness, headache, rash, hypertension, diabetes mellitus, joint pain, recent or past trauma, unconsciousness, tongue-bite and fecal/urinary incontinence, chest pain, palpitation, breathlessness, cough, sputum, hemoptysis, anorexia, nausea, diarrhea, constipation, vomiting, and heat or cold intolerance.After his convulsions were controlled, he noticed weakness on the left side of his body. The family members gave a history of similar but milder self-limiting episodes once or twice a year since he was a one-year-old. He was born to non-consanguine parents, had an uneventful birth history, through a breech presentation though, and was fully immunized against six communicable diseases as per the national schedule during childhood. He normally grew up to adulthood and his family neither ever reported to a doctor nor was he prescribed with any medication for his convulsions. None of his siblings had a similar symptom. He was a non-smoker and was not a substance abuser.

On physical examination, there was a port-wine stain (PWS) birthmark on the face, extending from the right supra-orbital ridge, right upper eyelid, and right side of the nasal bridge to right ala nasi (Figure 1). He had no anemia, jaundice, cyanosis, purpura, clubbing, koilonychia, leuconychia, edema, or dehydration. His thyroid gland was not enlarged and lymph nodes were not palpable. His pulse was 89/minute, regular, with normal volume and character. His blood pressure was 120/80 mmHg; his respiratory rate was 15 breaths/minute, his core temperature was 98°F, and his jugular venous pulse was normal. He had normal higher psychic function with an Abbreviated Mental Test-10 score of 9 and normal speech. There was no cranial nerve palsy and the sensory system was intact. Motor examination revealed upper motor neuron type weakness in the left upper and lower limbs, increased tone, diminished power (4/5), exaggerated tendon reflexes, and extensor plantar response on the side of weakness, and there was mild wasting in the left lower limb (Figure 2). He had no extra-pyramidal, cerebellar, or dysautonomic signs but had a mild form of a hemiplegic gait. His precordium, chest, abdomen, and locomotor system revealed unremarkable physical findings. A thorough ophthalmologic evaluation including fundoscopy, slit-lamp examination, and tonometric tests revealed normal findings.

Routine blood tests including complete blood counts, peripheral blood smear, blood glucose, serum creatinine, electrolytes, calcium, bilirubin, total protein, albumin, globulin, liver enzymes, and prothrombin time were within normal ranges. His chest radiograph was normal, but an X-ray skull showed calcification in the right parieto-occipital region (Figure 3). Axial computed tomographic scan of the brain revealed diffuse calcification involving predominantly subcortical white matter of the right cerebral hemisphere in the fronto-parieto-occipital regions. The right lateral ventricle was slightly dilated due to volume shrinkage of the right hemisphere and a mild form of right cerebral atrophy was also noted. But no significant midline shift was seen, and sella, parasellar areas, and posterior cranial fossa were normal (Figure 4). The patient's seizures were controlled with anticonvulsant medications.

A presentation with recurrent seizure, commencing in childhood in presence of ipsilateral facial PWS and radiological evidence of ipsilateral cerebral calcification with normal ocular findings comprised the diagnosis of Sturge-Weber Syndrome Roach Type I.

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FIGURES

Figure 1: Port-wine stain on the face

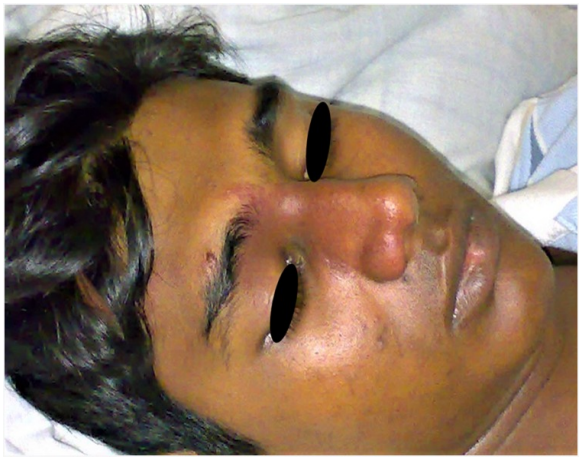


Figure 2: Wasting of left lower limb

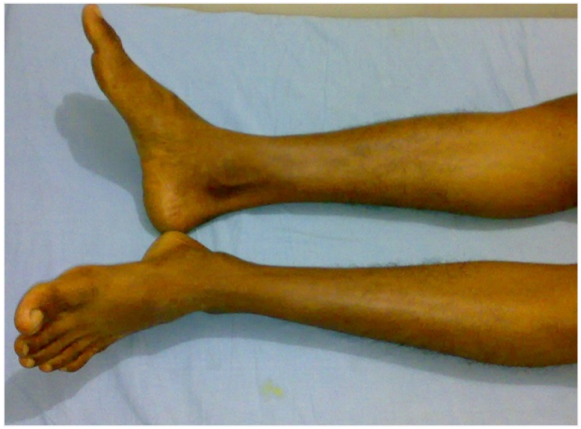


Figure 3: Calcification on skull X-ray (arrow)

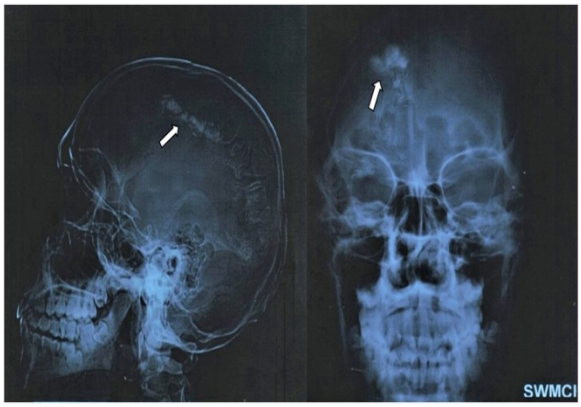
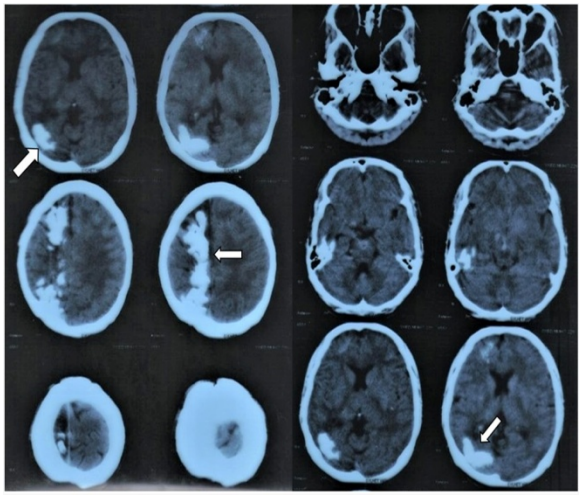


Figure 4: Subcortical cerebral calcification on CT head (arrow)



Discussion

Sturge-Weber Syndrome (SWS), a rare congenital angiomatosis, simultaneously involving the skin, mucous membrane, brain, and/or eyes, gives rise to a neuro-cutaneous syndrome of variable clinical presentation, resulting from mutation of the GNAQ gene.[4,5] The clinical features are often categorized to type I, type II, and type III according to the Roach scale (Table 1). Type I is called the classical type and Type III is the rarest type.[8]This report presents a 22-year-old man with SWS type I without any ocular problem. In different studies, it has been seen, that SWS has a male preponderance, and the age of presentation ranges from birth to the second decade of life, and the majority of them present before the second birthday.[5,6,9]

Capillary hemangioma of the face known as port-wine stain (PWS) is one of the common features of SWS seen in types I and II, distributed along the branches of the 5th cranial nerve. They are sometimes called nevus flammeus, are usually unilateral, and more commonly along the ophthalmic division of the trigeminal nerve. It can sometimes be bilateral and may extend beyond the face to neck and extremities, according to some reports.[10] Initially it is deep red but darkens with increasing age and may bleed on trauma.[11] Some cases present to the dentistry facilities with bleeding from gingival angiomas, and some have angiomas in

the lips, palate, or beneath the tongue. The involvement of structures in the oral cavity imposes increased risk but is less frequently encountered. [12]

The neurological manifestations are due to angiomatosis in the lepto-meninges which leads to cerebral ischemia. Chronic ischemia leads to brain atrophy, replacement of damaged area by glial tissue, and persistent deposition of calcium salts in the affected areas. The clinical and radiological features are produced by these pathological changes in the brain.[12,13] Unilateral leptomeningeal lesions are more common but bilateral lesions have also been reported.[7,10] Cerebral ischemia causes convulsions, and sometimes hemiparesis, whereas cerebral atrophy causes progressive mental retardation in half of the cases. One-third of cases give a history of vascular headaches. Convulsions manifesting in early childhood, are the commonest neurological feature of SWS, which are usually focal complex or secondary generalized partial in type.[14]The index case presented with convulsions from one year of age with new onset of left hemiparesis in adulthood. However, his mental scores were normal, and had no developmental delay or headache. He had a PWS in the distribution area of the ophthalmic branch of the right trigeminal without any oral extension, and his ocular findings were unremarkable.

A number of ocular problems have been reported in SWS cases, including nevoid marks in the sclera, conjunctival telangiectasia, bupthalmos, choroidal hemangioma, and glaucoma, arterio-venous angiomas in the optic nerve, and aneurysms in retinal vessels.[5,9] Vascular proliferation in the trabecular meshwork of the eyeball leads to the development of a very difficult category of glaucoma for ophthalmologists to treat. In a study, Helmi et al found that around seventy percent of ocular cases of SWS had glaucoma. Though glaucoma is common among SWS cases, its absence does not rule out a diagnosis.

Recent case reports represent increasing evidence of endocrine, otolaryngological, and psychiatric manifestations in SWS. Endocrine dysfunctions in the form of growth hormone insufficiency and secondary hypothyroidism have been reported.[13,14]Medical treatment of seizures has widely been adopted as a means of seizure control, but some patients continue to have intractable convulsions in spite of optimum treatment. In them, surgery is a presumed alternate. However, a case-based systematic review and meta-analysis failed to demonstrate any significant improvement in seizure control after lesionectomy in SWS patients; but post-operative cases showed improvement in hemiparesis.[15]

Table 1: The Roach scale

Type of SWS	Leptomeningeal angioma	Facial angioma	Glaucoma
Type I	Present	Present	Present or absent
Type II	Absent	Present	Present or absent
Type III	Present	Absent	Usually absent

The presence of brain calcification provides important evidence of leptomeningeal angiomas, and tissue diagnosis is usually not warranted. Central nervous system calcification in SWS conforms to the sulci and gyri and often gives rise to the pathognomonic radiological feature of cortical tram-track or rail-road sign. Calcifications can be visualized in plain skull X-ray films but computed tomographic scans reveal diagnosis in most cases of SWS with seizures at presentation. However, at present, gadolinium contrast magnetic resonance imaging is the investigation of choice for SWS diagnosis.[16]Characteristic electroencephalographic (EEG) features are asymmetric, low voltage discharge in focal areas that change with the advancing age of the patient.[7,8] The changing pattern of perfusion scan from hyper-perfusion to hypo-perfusion over time indicates a progressive decline of brain function in SWS. Cerebral angiography is not a routine test for SWS but can be of help to exclude other forms of congenital vascular abnormalities in non-classical cases.[10,11,12]

Conclusion

Sturge-Weber syndrome is a rare neuro-cutaneous disease that presents with seizures in more than eighty percent of cases. In patients who present at an adult age, careful past history from childhood, a search for angiomas in the

face, neck, or limbs, and demonstration of brain calcification in X-ray or magnetic resonance images can lead to an easy diagnosis. Though this case had classical features, it should be borne in mind that presentation can be atypical due to the involvement of unusual sites.

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