

Sturge Weber Syndrome - A Case Report with Literature Review

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Abstract

Sturge Weber syndrome (SWS) is a rare neurocutaneous disorder. It is characterized by the abnormalities affecting the brain (pial angiomas), skin (facial port-wine birthmark) and eyes (commonly) glaucoma. It is a congenital, non-familial and sporadic condition associated with somatic mutation in *GNAQ* gene. Symptoms report varies in severity and age of onset, but it is commonly detected in infancy. This case illustration is that of a 4-year old who presented with fever and focal seizure with secondary generalization. Examination revealed quadriplegia, more marked in the right lower limb. Brain computerized tomography (CT) scan showed gyral calcification, atrophy of the cerebral hemisphere and early cerebritis. She was commenced on anti-epileptic drugs (AED), antibiotics and supportive care.

Keywords: Sturge-Weber Syndrome; Facial Naevus; Seizures; Anticonvulsant

Introduction

Sturge-Weber Syndrome (SWS) is a congenital, non-hereditary disorder affecting the brain, eyes and skin. It is also known as encephalotrigeminal angiomas. The first case was described by Schirmer who did not recognize it was neurological disorder. In 1879, William Allen Sturge gave a more precise description of the disease and associated the dermatological and ophthalmic changes of the disease to neurologic symptoms. Frederick Parkes Weber, in 1929, elucidated the radiologic features seen in patients of SWS [1].

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 [2]. Neurocutaneous syndrome or phakomatoses are disorders characterized by abnormal development of growths in the skin, central nervous system (CNS), bones and other organs. SWS, for example, show pial angioma, facial port wine stain and ocular abnormalities.

It is classified using the Roach scale [1] into:

Type 1: Presence of both facial and leptomeningeal angiomas; there may be associated glaucoma.

Type 2: Presence of facial angiomas alone; there may be associated glaucoma.

Type 3: Isolated leptomeningeal angiomas; usually no glaucoma.

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 anti-epileptic drugs. Surgical resection is reserved for intractable cases of seizures.

Case Illustration

A 4-year old female being managed for delayed development milestones who presented with high grade fever of ten days duration and focal tonic-chronic seizure with secondary generalization. There was associated headache but no history suggestive of meningism, though had history suggestive of tonsillopharyngitis.

Examination findings on presentation revealed a female pre-school child who had a reddish patch on the right side of the forehead

in the distribution of ophthalmic division of trigeminal nerve. She was unconscious with Glasgow Coma Score of 11/15. She had limb weakness, more marked on the right lower limb with muscle power of 2/5 versus 4/5 on the left and plantarflexion on the left ankle. There was hypertonia and hyper-reflexia on the right lower limb.

Brain CT scan revealed right cerebral and gyral calcification with ipsilateral atrophy and left focal parietal early staged cerebritis.

She was commenced on intranasal oxygen, antibiotics antipyretic and AED (loading and maintenance). she became seizure free and gradually regained consciousness. She was discharged home on oral antibiotics and AED after two weeks on admission. She has had two clinic visit and still remaining seizure free on AED. (Figure 1).

Discussion

Sturge-Weber syndrome is congenital, non-familial and sporadic developmental disorder. Its etiology is poorly understood but studies have shown that it is caused by somatic mutations in the *GNAQ* gene, located on the long arm of chromosome 9 [3,4]. It is a rare disorder. The exact incidence is unknown but estimated to be 1 in 20,000-50,000 live births [2]. It is cosmopolitan with no racial predilection and equal occurrence in males and females [1].

The exact pathogenesis is not fully understood but theories exist. It has been postulated that the occurrence of leptomeningeal and facial angiomas suggest the persistence of undeveloped sinusoidal vascular channels. Another theory is the incomplete development of superficial venous drainage with subsequent compensatory dilatation of small venous channels [2]. This relative deficiency of superficial cortical veins, there is shunting (steal) of blood to the deep venous system by the enlarged medullary veins with resultant stasis and ischemia, eventually

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[2]. Ultimately, these changes culminate in epileptic convulsive crisis, transient hemiparesis, gliosis, and progressive deposition of calcium salts same in above case illustration. The cortical calcification impresses the characteristic double contoured “tram-line” appearance, a finding seen in above CT scan. This can be readily appreciated with radiological imaging studies. Brush field and Wyatt stated that these tram-line calcifications are pathognomonic of SWS [4]. The characteristic facial nevus is composed of multiple thin-walled vessels that are similar to capillaries.

Recent studies have shown that SWS is caused by a somatic mutation in a nucleotide transition in the gene *GNAQ* on the long arm of chromosome 9 (specifically 9q21). This mutation results in increased activity in pathways transmitting signals from a subset of G protein coupled receptors (GCPR) [3,4]. The exact mechanism by which this activation lead to port-wine birthmarks and SWS is not well known, as such additional studies is required [4].

The clinical features of SWS can be neurological and or non-neurological. The neurological symptoms include seizures, hemiparesis, headaches, visual field deficits, cognitive impairments as seen in above case illustration. Other features include early handedness, gaze preferences as well as stroke-like episodes [4]. The non-neurological symptoms of SWS include behavioral and emotional derangement, endocrine problems, learning difficulties, and other medical issues [1].

The first manifestation is usually a seizure and in 90% of patients, it occurs in the first year of life. They may be in the form of infantile spasms, myoclonic, atonic or tonic seizures. The seizures may be focal or generalized. It has been observed that seizures are often precipitated by extrinsic factors such as lack of sleep, stress, and illness (including infection) and are mainly thought to arise from epileptic focus caused by calcification [4]. These seizures can be associated with other neurological symptoms, commonly hemiparesis and delayed developmental milestones. Initially, it is responsive to AED, but they may progressively become recalcitrant to anticonvulsants and thus become an indication for surgery [5].

Cutaneous manifestation is common presentation in SWS, notably facial nevus, usually in the distribution of ophthalmic and maxillary divisions of the trigeminal nerve. In the above case, it was in the ophthalmic division. SWS is typically unilateral. It is present at birth, remaining static throughout life [5]. This is in contrast to the infantile hemangioma which is not present at birth. However, it is more common than port-wine stain. Facial port wine stain is associated with a 10% to 35% risk of SWS. However, they are not pathognomonic of SWS [4]. Apart from the brain and skin, SWS also commonly affects the eyes with glaucoma being the commonest ocular manifestation. When present, it is almost always ipsilateral to the facial port-wine stain. Other Ocular manifestation include buphthalmos, increased vascularity of the conjunctiva, increased tearing and strabismus [2].

Diagnosis of Sturge-Weber syndrome is based on the presence typical clinical manifestation including neurological symptoms and cutaneous appearance, supported by imaging findings on brain MRI and or CT scan [2], similar pattern of arriving at diagnosis of SWS in above index case.. SWS can also be diagnosed in the presence of two out of three of the following: increased ocular pressure, facial nevus, and leptomeningial angiomatosis. Patients with leptomeningial angiomatosis, without skin or eye involvement, are qualified as having the intracranial variant of SWS [1].

Magnetic resonance imaging (MRI) scan is the preferred imaging modality for diagnosing and monitoring brain involvement, especially

in patients above the age of one. The recommended standard is a T1 and T2-weighted brain MRI scan with gadolinium contrast, and post-contrast fluid attenuated inversion recovery (FLAIR) [1], however, this was not done for the index case as it is unavailable here in our rural settings

Gyriform calcification is a common feature that is appreciated on the skull radiographs and classically described as “tram-track sign” [6]. The best modality to detect calcifications is CT scan and was described in above displayed CT scan in Figure 2. It can also show the other changes such as cortical atrophy and lepto-meningeal enhancement on the post-contrast studies. However, the use of ionizing radiations is a major drawback with the use CT scan. Therefore, contrast MRI scan is the recommended imaging modality of choice [2].

Other investigation modalities include Electroencephalography (EEG) to assess brain function and ocular ultrasound to detect ocular abnormalities. FDG-PET and MR spectroscopy have also found roles in the evaluation of patients with SWS. They can be used to help screen surgical candidates, or as research tools. They are currently not used as routine clinical tests for diagnosis or monitoring disease severity [4] and above investigations were not done for the patient in view of its unavailability.



Figure 1: Showing facial nevus in V1 distribution.

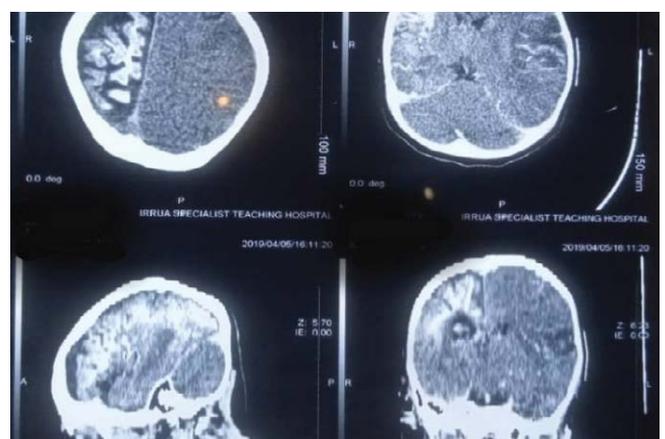


Figure 2: Showing intracerebral calcification with ipsilateral atrophy and contralateral cerebritis.

Treatment is generally supportive and primarily aims to minimize seizures activity with anti-epileptic medications. Surgery may be offered to patients whose seizures are refractory to available medical therapies [2]. Therefore, the mainstay of treatment is the control of seizures with the use of anticonvulsants. Treatment should ideally start after the first focal seizure and the most common first line anticonvulsant is oxcarbazepine [4]. This is however, scarce in our environment. Other anticonvulsants to with proven efficacy include levetiracetam and topiramate. They can also be used as first line in the absence of oxcarbazepine [4]. Potentially effective, but less commonly used chronic anticonvulsants include valproic acid, carbamazepine, zonisamide, lamotrigine, and phenobarbital which was used for this patient.

It is also essential to mitigate precipitants of seizures such as stress, lack of sleep, and illness. As in our reported case, when patients do get sick, control of fever, treatment of infection and proper hydration are an essential part of management. Cerebritis as seen in this patient is not commonly associated with SWS and it was a coincidental finding which probably precipitated the epileptic attack.

The recent years has seen trial of newer treatment measures aimed at preventing seizures and getting better control. Low-dose aspirin has been offered to a small group of infants and young children prior to the onset of seizures or strokes with varying outcomes [7]. More studies will be necessary to ascertain the efficacy of aspirin in seizure prophylaxis. The Modified Atkins Diet (MAD) may be useful in controlling seizure activity [4].

Up to 50% of patients achieve seizure control with use of medications. With newer anticonvulsants and low-dose aspirin this percentage is probably higher [7]. For the rest patients, lifestyle changes and avoidance of seizure precipitants are important, with surgery being a final resort [4]. Surgery is reserved for patients with severe symptoms that are not responsive to medical therapy. Examples of available surgical procedures syndrome include hemispherectomy or focal resection of the seizure focus. It is contraindicated in patients with bilateral involvement [2].

For glaucoma, the first line of therapy is with use of topical medication, although it is often insufficient. The commonest medication in use is latanoprost, which has been shown to provide effective control of intraocular pressure in up to 50% of patients [4]. Ocular surgery may be necessary when glaucoma is recalcitrant to medications. Surgical

options include trabeculotomy, trabeculectomy and goniotomy in infants and children. Others include non-penetrating sclerectomy, the use of valve drainage implants, and cilio-destructive procedures in adults [8].

For the facial port wine stain, laser treatment can result in lightening of the port-wine birthmark [4]. There are many differential diagnoses of Sturge-Weber syndrome. These include, among others, Blue Rubber Bleb Nevus Syndrome, Klippel-Trenaunay-Weber Syndrome, Wyburg-Mason syndrome, and PHACES (posterior fossa abnormalities, hemangiomas, arterial anomalies, cardiac, eye, and sternal anomalies) [2].

Conclusion

Sturge Weber Syndrome is a rare congenital disease which has classical symptomatology and neurological imaging findings. The treatment goal is symptom driven and involves multi-specialists including the Neurosurgeon, Neurophysician, Ophthalmologist, Cosmetologist, Physiotherapist, and radiologist. Treatment options include pharmacological, surgery and laser therapy. The place of surgery in refractory seizure or progressive disease is well documented but more studies will be required to establish the place of surgery in the management of early disease.

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