

Sturge Weber Syndrome Type III – A Rare Case Report

¹A. C. Mammen, ²S.K. Murmu, ³S.C. Majhi, ⁴D. Sethi, ⁵B. Deepti.

From the department of Pediatrics, VSS Medical College, Burla, Sambalpur, Orissa.

Abstract: *Sturge Weber angiomas is a rare, nonhereditary developmental condition characterized by a hamartomatous vascular proliferation involving the tissues of brain and face. A report of a case with no facial port wine stains, and only CNs abnormality is described.*

Keywords: *sturge weber syndrome III, neurocutaneous markers, convulsion.*

I. Introduction

Sturge weber syndrome (encephalotrigeminal angiomas) is a congenital, non-familial disorder of unknown cause. It is characterized by

- i. A facial port-wine stain affecting the facial skin (in the distribution of V1 & V2 division of trigeminal nerve.
- ii. Vascular eye abnormalities
- iii. An ipsilateral occipital leptomenigeal angioma.
- iv. It may also consists of congenital hamartomatous malformations that may affect the eye, skin, and central nervous system (CNS) at different times, characterized by the combination of venous angiomas of leptomeninges, face, jaws and oral soft tissues.[5] SWS was first described by Schirmer in 1860. More specific description was given by Sturge in 1879.[6]
- v. SWS is believed to be caused by the persistence of vascular plexus around the cephalic portion of the neural tube. This plexus develops during the sixth week of I.U. development but normally undergoes regression during ninth week.[7]
- vi. Angiomas of leptomeninges are usually unilateral, located in parietal and occipital region. The presence of angioma results in alteration of vascular dynamics causing precipitation of calcium deposits in cerebral cortex underlying the angioma. Seizures, mental retardation, hemiplegia, or hemiparesis may develop secondary to this and their severity depends on the extent of lesion.[8]
- vii. The cutaneous angiomas are called port wine stains, which usually occur unilaterally along dermatomes supplied by the ophthalmic and maxillary division of trigeminal nerve. It may be bilateral or totally absent or may extend to neck, limbs and other parts of the body.[8] Involvement of the area supplied by ophthalmic division is pathognomic. Ocular involvement can result in glaucoma, choroidal hemangioma, bupthalmos, or hemianopis.[9]

Case Report: An 8 year old male child belonging to lower socioeconomic status, came to our department with history of one episode of focal convulsion involving the right side of the body, unprovoked, lasting for 3-4 minutes, associated with no loss of consciousness or post ictal stage. It was not associated with any fever, vomiting, sleep deprivation or any drug intake, CSOM, rash, rhinitis, or alcohol intake. There was no prior history of any seizure, contact with open case of TB. There was no family history of any seizure disorders, sickling or siblings with developmental delay. Patient was immunized according to NIS and perinatal, dietary and developmental history was normal.

On examination, child was moderately built and nourished, afebrile. There was nopallor, icterus, cyanosis, clubbing, oedema or lymphadenopathy. There was no evidence of neurocutaneous markers (Fig 1) or rash. He weighed 20kg, and height was 115cm. Systemic examination revealed cardiovascular, respiratory and gastrointestinal system within normal limits. CNS examination revealed, child being conscious, oriented to time place and person, memory, speech, intelligence being normal. Sensory system examination revealed no decreased sensation in any of the body parts. Motor system examination revealed grade 2/5 of the right upper limb and lower limb while the left side revealed grade 5/5. Reflexes were exaggerated on the right side when compared with the left side. There was no difference of tone in both sides of the body. There were no cerebellar or meningeal signs. Brain and spinal cord examination was normal.

Patient was treated with anticonvulsant and was on i.v fluids and physiotherapy was advised. Blood panel was sent and all were normal. Ophthalmological examination was normal. CT & MRI revealed left occipital meningeal angiomas with calcifications (Fig 2 and 3), suggesting a diagnosis of SWS Type III. The child was discharged on the 5th day with oral anticonvulsants and physiotherapy was advised to continue back at home.

II. Discussion

SWS is a non-familial congenital disorder of unknown incidence and cause. According to National Institute of Neurological Disorders and Stroke, SWS is a neurological disorder. It is characterized by a congenital facial birthmark, known as a capillary malformation and other neurological abnormalities. The other symptoms include eye and internal organ irregularities which vary in degree [1].

The most apparent sign of SWS is a birthmark or port wine stain (PWS) on the face. The PWS is due to an overabundance of capillaries just beneath the surface of the involved areas. When the port wine stain covers the eye and forehead region of the face, SWS should be considered. In rare instances, SWS is present even in the absence of the PWS. SWS consists of three basic types involving a PWS on the face, eye or the brain [2]:

Type 1, the most common, involves both facial and brain vascular malformations and may involve glaucoma. Seizures usually occur within the first year of life as a result of the brain involvement. Eye involvement is normally noted within the first year of life as well. The white portion of the eye may appear bloodshot due to the overgrowth of blood vessels on the eye.

In rare cases, the facial and brain aspects involve both sides of the head. Mental and physical development can be impaired to varying degrees, depending on the amount of vascular birthmark throughout the brain and eye.

Type 2 involves a vascular malformation on the face and the possibility of glaucoma, but no evidence of brain involvement [3]. Recognizing the facial port wine stain is often the first sign. Throughout the life of the individual, connected symptoms may include glaucoma, cerebral (brain) blood flow abnormalities, headaches and various other complications.

Type 3 is commonly noted to have vascular malformation in the brain, with no face stains and usually no development of glaucoma. Commonly referred to as forme fruste, this type is identified through brain scans with the use of a contrasting agent (special dye). It can be confused with other diagnoses prior to a brain scan and can be difficult to diagnose.

Out of the 3 types of sturge weber syndrome (Roach Scale)[4], only 24 cases of SWS Type III have been reported so far in literature.

Fig 1



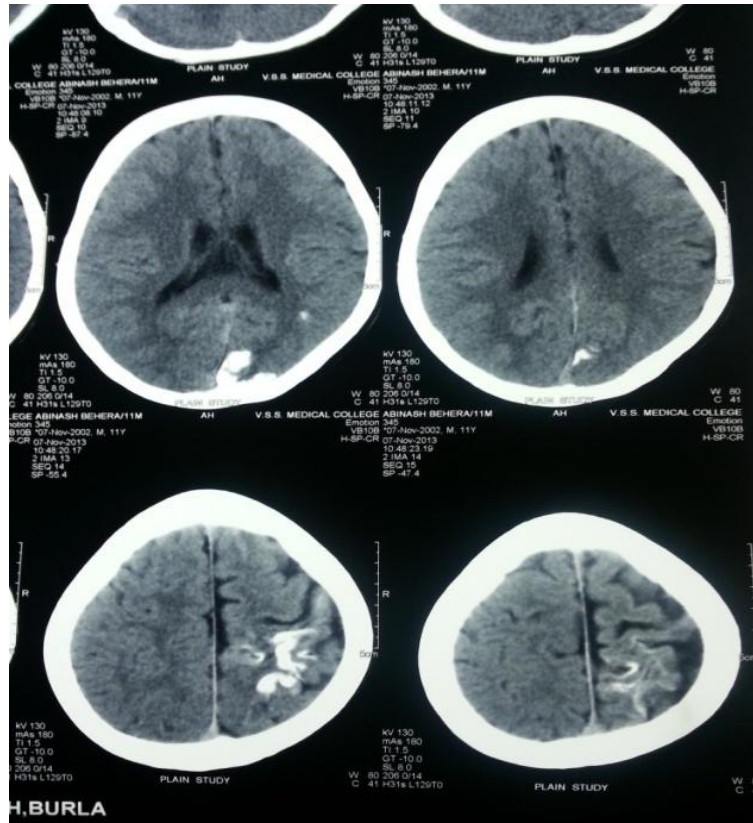


Fig 2

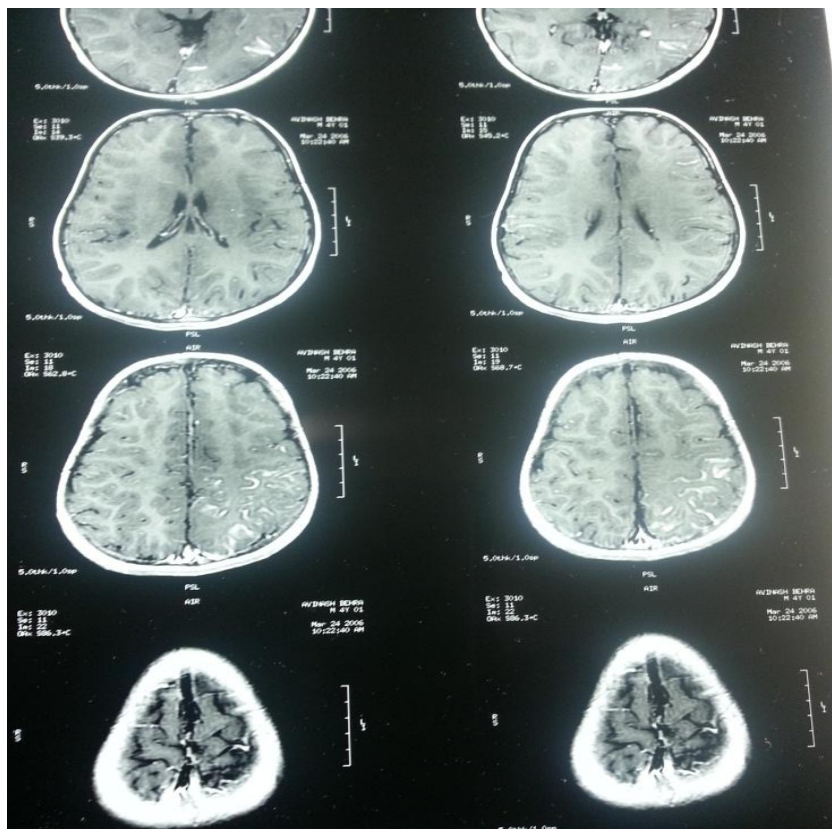


Fig 3

Funding: None
Competing interests: None stated

References

- [1]. Chaundhary SC, Sonkar SK, KumarV, GolchhaS, Sturge Weber Syndrome, J Associ Physicians India, 2011 May;59:327-9 (Abstract)
- [2]. The Sturge Weber Foundation: <http://www.sturge-weber.org>
- [3]. National Institute of Neurological Disorders and Stroke: http://www.ninds.nih.gov/disorders/sturge_weber/sturge_weber.htm
- [4]. <http://www.seattlechildrens.org/medical-conditions/common-childhood-conditions/sturge-weber/>
- [5]. <http://www.sturge-weber.org/medical-matters/sturge-weber-syndrome.htm>
- [6]. Kliegman, Stanton, St. Geme, Schor, Berhman, Sturge Weber syndrome, Nelson textbook of pediatrics, 19th ed pg 2051-2052
- [7]. Rajendran R. Benign and malignant tumors of oral cavity. In: Rajendran R, Sivapathasundharam B, editors. Shafer's Textbook of Oral Pathology. 6th ed. Noida: Elsevier; 2009. pp. 144–5.
- [8]. Caiazzo A, Mehra P, Papageorge MB. The use of preoperative percutaneous transcatheter vascular occlusive therapy in the management of Sturge–Weber syndrome – Report of a case. J Oral Maxillofac Surg. 1998;56:775–8. [PubMed]
- [9]. Neville BW, Damm DD, Allen CM, Bouquot JE, editors. 3rd ed. St. Louis: Elsevier; 2009. Oral and Maxillofacial Pathology.
- [10]. Suprabha B, Baliga S. Total oral rehabilitation in a patient with port wine stains. J Indian Soc Pedod Prev Dent. 2005;23:99–102. [PubMed]
- [11]. Gorlin RJ, Pindborg JJ. New York: McGraw- Hill; 1964. Syndromes of head and neck; pp. 406–9.