International Journal of Contemporary Pediatrics
Ananya SLT et al. Int J Contemp Pediatr. 2017 Nov;4(6):2220-2222
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: http://dx.doi.org/10.18203/2349-3291.ijcp20174760

Case Report

Encephalotrigeminal angiomatosis: an atypical presentation of a rare disease

Sai Lakshmi Ananya T.*, Radha Kumar

Department of Pediatrics, Saveetha Medical College and Hospital, Chennai, Tamil Nadu, India

Received: 29 September 2017
Accepted: 13 October 2017

***Correspondence:**
Dr. Sai Lakshmi Ananya T.,
E-mail: ananyatenali@gmail.com

ABSTRACT

Encephalotrigeminal angiomatosis also called as Sturge Weber Syndrome (SWS) is a sporadically occurring rare congenital neuro cutaneous disorder. Unilateral facial portwine stain along with contralateral focal seizures and ipsilateral leptomeningeal angiomatosis is considered as diagnostic of Sturge Weber Syndrome. Capillary malformation along the ophthalmic and maxillary divisions of the trigeminal nerve is common in this condition. MRI with contrast is the imaging modality of choice for the diagnosis. The current report describes a case of a 9-month-old infant with atypical presentation of Type 1 Encephalo-trigeminal angiomatosis with complications. The clinicopathological and radiographic features and differential diagnosis are discussed.

Keywords: Angioma, Atypical presentation, Portwine stain, Roach scale, Sturge Weber syndrome

INTRODUCTION

Encephalo trigeminal angiomatosis was first described by Schirmer in 1860 and subsequently more specific description was given by Sturge in 1879.[1]

It is also called as Sturge Weber syndrome, leptomeningeal angiomatosis, Sturge-Kalischer-Weber syndrome or leptomeningofacial angiomatosis.[2]

Sturge Weber syndrome is a sporadic vascular disorder, a rare neurocutaneous syndrome with incidence of 1:50,000 live births presenting with portwine stain, leptomeningeal angioma ,glaucoma due to capillary malformation and abnormal blood vessels.[3]

Treatment and prognosis depends upon the nature and severity of clinical features and associated complications. In this case report, an atypical case of Type 1 Encephalo trigeminal angiomatosis with complications is discussed.

CASE REPORT

A 9-month-old infant was admitted with the chief complaints of red patches on the face and chest which was present from birth with recurrent focal seizures (Figure 1).

He was the first born of non-consanguineous marriage with no significant antenatal and family history. The baby was delivered at term by normal vaginal delivery with a birth weight of 2.7 kg. The child was apparently normal till 3rd month of life when he developed recurrent focal seizures involving the right upper and lower limb for which he was receiving multiple antiepileptic drugs such as levetiracetam, phenytoin sodium, sodium valproate and clonazepam. The seizures were only partially controlled with the medications inspite of good drug compliance. There was history of right sided weakness involving right upper and lower limb for fourth month of life.

The child had history of global developmental delay affecting all the domains including delay in language and social development. At the 9th month of age, the child had attained partial neck holding and palmar grasp. He had early left-hand preference. On examination, port wine stain was seen over the entire forehead and both cheeks, right side of the neck and shoulder and extending over the right side of the trunk, right hand and foot. His weight was 6.5 kg and head circumference was 41 cm. There was hemi hypertrophy involving right upper and lower limbs. The child had right sided hemi paresis with hypertonia and brisk deep tendon reflexes. There were no dysmorphic facial features. Ophthalmologic examination showed glaucoma and megalocornea of left eye. On radiographic investigations, CT and MRI brain scan revealed bilateral cerebral atrophy which was more on the left side of the brain (Figure 2 and 3).

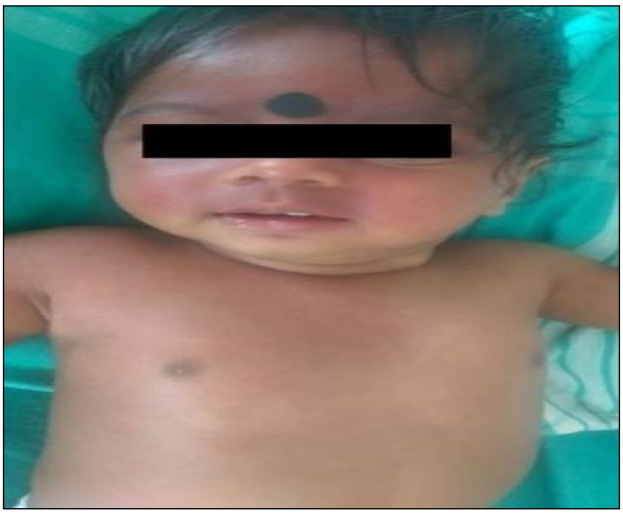

Figure 1: Portwine stain seen involving the face bilaterally, right side of neck and trunk.

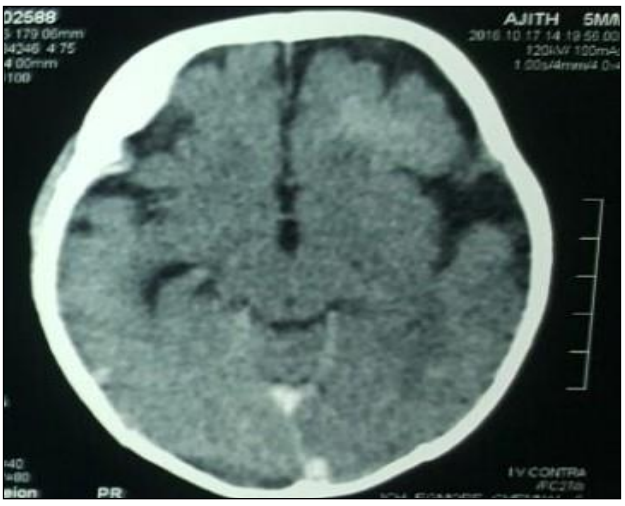

Figure 2: CT with contrast showing bilateral cerebral atrophy more on the left side.

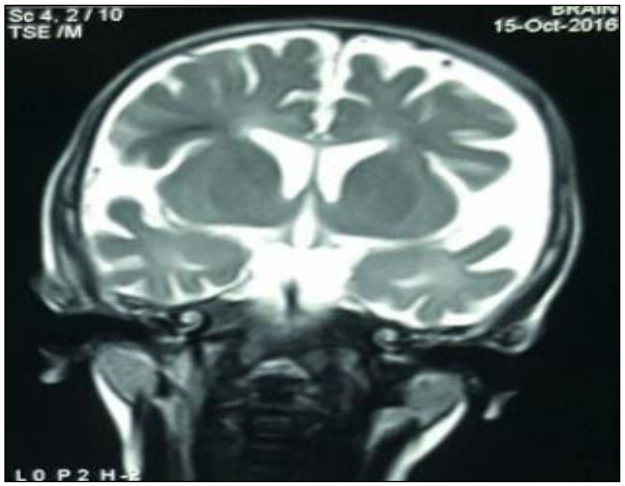

Figure 3: MRI brain showing left sided cortical atrophy seen.

Abnormal EEG activity was noted in left parietal lobe. Based on the clinical presentation of portwine stain, focal seizures, glaucoma and neuro imaging studies, diagnosis of Sturge-Weber syndrome (SWS) was established. According to ROACH scale, the child was diagnosed as Type I SWS. After seeking neurological and ophthalmology opinions, management of the child was planned. The parents were counseled regarding progression of the disease, management options and prognosis. The dosages of anti-epileptics were increased after which the episodes of seizures decreased markedly. Medical management for glaucoma was initiated and baby is on regular follow up.

DISCUSSION

Sturge Weber syndrome is a rare neuro cutaneous syndrome presenting with vascular predominance and is grouped under phakomatoses. The incidence of SWS is estimated at 1 per 50,000 live births.[3] There is no regional or racial predominance and it affects both the sex equally.[4] It is characterized by unilateral facial cutaneous vascular malformation which is called nevus flammeus or port-wine stain along with ipsilateral leptomeningeal angiomatosis. The basic pathology is persistence of vascular plexus around the cephalic portion of the neural tube which develops during the sixth week and undergoes regression normally during ninth week of intra uterine gestational age.[4] These malformations lead to venous hypertension and subsequent hypoperfusion of the underlying cortex causing chronic cerebral ischemia, atrophy, and neurological deterioration. The genetic basis for SWS has been recently found to be somatic mutation in GNAQ gene.[5] Port wine stains typically occur unilaterally along the dermatomes supplied by the ophthalmic and maxillary division of trigeminal nerve. Ocular association can result in glaucoma, choroidal hemangioma, bupthalmos, or hemianopis.

Port wine stains should be differentiated from nevus flammeus neonatorum that occurs as a secluded lesion, pyogenic granuloma and venous varicosities.[6] Differential diagnosis of Sturge Weber syndrome includes Klippel Trenauny-Weber syndrome, hereditary hemorrhagic telengiactasias, Von Hippel Lindau disease.[1]

Roach scale is used for classification of SWS as Type I (both facial and leptomeningeal angiomas present; glaucoma may be present), Type II (only facial angiomas present; glaucoma may be present) and Type III (isolated leptomeningeal angiomas; usually no glaucoma).[3]

The infant had atypical features of SWS as he had bilateral portwine stains which is seen in less than 33% of cases.[7] Port wine stains can be improved by dermabrasion and flash lamp pulsed dye lasers. Topical sirolimus is now being used in combination with laser therapy to prevent re-growth of abnormal vessels.

Seizures are treated with anti-epileptic drugs sometimes in combinations and even then, not all patients respond completely to medical management (refractory seizures). In such patients, surgical techniques like hemispherectomy, focal cortical resection, and vagal nerve stimulation are helpful. Complications like glaucoma are managed medically with eye drops (beta antagonists, anti cholinergics) but ultimately surgical intervention is required.

CONCLUSION

There is a greater likelihood of intellectual impairment when seizures start before the age of 2 years and in seizures resistant to treatment. Prognosis is worst in the minority of children who have both sides of the brain affected by the blood vessel abnormalities.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Gill NC, Bhaskar N. Sturge-Weber syndrome: a case report. Contemp Clin Dent. 2010;1(3):183-5.
2. National Organization for Rare Disorders (NORD). Rare disease information: Sturge Weber syndrome. 2017. Available at https://rarediseases.org/rare-diseases/sturge-weber-syndrome/ Accessed on 26 September 2017
3. Neurocutaneous syndromes. In: Kliegman RM. Nelson textbook of pediatrics. 20th edition. International Edition Elsevier; 2016:2876-7.
4. Sharma N, Passi S, Mehta A. Sturge-Weber syndrome: report of a case and literature review. J Pediatr Dentistry. 2014;2(2):65.
5. Shirle MD, Tang H, Gallione CJ, Baugher JD, Frelin LP, Cohen B, et al. Sturge-Weber syndrome and Port-Wine stains caused by somatic mutation in GNAQ. N Engl J Med. 2013;368:1971-9.
6. Neto FXP, Junior MAV, Ximenes LS, Jacob CCS, Junior AGR, Palheta CP, et al. Clinical features 2. of Sturge-Weber syndrome. Arq Int Otorrinolaringol. 2008;12:565-70.
7. Ahmed K, Yaseen A, Hassan I, Shah PA. Sturge-Weber syndrome with bilateral nevus flammeus. Indian J Pediatr Dermatol. 2013:14(3):79-82.

Cite this article as: Ananya SLT, Kumar R. Encephalotrigeminal angiomatosis: an atypical presentation of a rare disease. Int J Contemp Pediatr 2017;4:2220-2.