

Case Report on Sturge-Weber syndrome

Vyshnavi Kurra*, Ramya Bala Prabha, Sushanta Kr Das, K. Abbulu

Department of Pharm D, CMR College of Pharmacy, Hyderabad-501401, Telangana State, India

ABSTRACT

Sturge-Weber syndrome (SWS) belongs to a group of disorders collectively known as the phakomatoses. It 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. SWS is referred to as complete when both central nervous system and facial angiomas are present, and incomplete when only one area is affected. Epilepsy is the most common and often the first neurological complication of SWS. Here we present a case of seizure with hypocalcaemia to the risk of Sturge weber syndrome. Brain - MRI with contrast, however, be advised that due to the progressive nature of SWS, brain involvement may not be evident on early scans. So initially child was treated for seizures with anti-epileptic drugs. The health professionals have to be suitably able to recognize its characteristic signs and symptoms, and so improve the quality of life of the patients.

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vyshu.yadav@gmail.com

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Sturge Weber Syndrome (SWS) also known as encephalotrigeminal angiomatosis, is a rare sporadic non-familial neuro-cutaneous disorder that affects the meninges (most often the pia mater and arachnoid mater) of the brain and the skin of the face unilaterally. In rare cases the involvement may be bilateral [1]. Schirmer first described the association of a facial port wine stain (PWS) with bupthalmos in 1860. In 1879 Sturge described the association of a PWS with bupthalmos with focal seizures and postulated naevoid involvement of brain. The early radiological features of SWS were first reported by Weber 1922 and Dimitri 1923 [2]. SWS is referred to as complete when both central nervous system and facial angiomas are present, and incomplete when only one area is affected [3]. It occurs with a frequency of approximately 1 per 50,000. Males and females are equally affected and there is no racial predilection [4,5]. Sturge weber syndrome is caused due to somatic activating mutation occurring in the GNAQ gene [6]. Epilepsy is the most common and often the first neurological complication of SWS. Here we present a case of seizure with hypocalcaemia to the risk of Sturge weber syndrome.

Case report:

A 5month old female child was admitted in pediatrics department with chief complaints of seizures of 4-5 episodes, each episode with a gap of 2 hrs lasting for 5 to10 minutes with clonic movements of right upper

limb, deviation of mouth towards right and up rolling of eyeballs with non-responsiveness.

The child had history of fever simultaneous with onset of seizures and also known to be afebrile in between seizures (Febrile seizures). Initial episode of seizure (12.15 pm) was treated with Inj. Midazolam 0.5cc mixed in 0.5 cc normal saline by which the seizures are subsided. For another episode (2.30 pm) treatment with Inj. Midazolam 0.5 cc mixed with 1cc normal saline was given where the seizures continued so Inj. Phenytoin loading dose was given to control the seizures.

Birth history of the child revealed that the child was born out of 3rd degree consanguinous marriage with 3rd in order, birth weight was 1.8 kg, had history of delayed cry and was admitted in NICU on day 3 of life in v/o hypoglycemic seizures, CRP+ sepsis admitted for 15 days and improved by the time of discharge. She was given Inj. Augmentin, Inj. Amikacin, Inj. Meropenem for 7 days. Immunisation history was appropriate as per NIS. Ante natal history revealed that mother had severe preclampsia which was diagnosed in 3rd trimester. Developmental history of child include neck holding+.

On examination child was active, capillary hemangioma was present over the tip of nose on right side with a size of 2x3 cm which is progressively increasing in size since birth. Patient was found to be

conscious and coherent, afebrile, cardiovascular sound S1 & S2 heard, bilateral air entry was normal & no abnormal sound was present on respiratory examination, per abdomen was soft. CNS shows bulged anterior fontanelle with normal tone. Her laboratory reports shows normal study of complete blood picture except for serum calcium which was reduced. Upon admission she was prescribed with:

1. Inj. Phenytoin – 100mg (IV) in 30 cc normal saline over 30 mins at a rate of 20mg/kg.
2. Inj. Midazolam – 0.5 cc in 0.5 cc normal saline, IV, SOS.
3. Inj. Ceftriaxone - 250 mg IV, BD at a rate of 100mg/kg.
4. Anamol rectal suppository – 80 mg, PR, SOS.
5. IVF: Iso- P - 500ml/24hr at a rate of 20 ml/hr.

On day 2 (12-9-15) baby was moderately active, vitals were stable and was diagnosed as seizure with hypocalcaemia to the risk of sturge weber syndrome. Ultrasound pelvis scan was done which shows no sonographic abnormalities. To the existing treatment Syp. Paracetamol 3ml, PO, TID and Inj. Calcium gluconate 2ml/kg (10ml) in 10ml 5% dextrose over 30mins were added with an advice of heart rate monitoring. On day 3 (13-9-15) calcium gluconate and day 4 (14-9-15) IVF were stopped. On day 8 (18-9-15) MRI was performed which revealed an essentially normal study. Treatment was continued upto 12 days with an advice of watch for seizure. Later the patient was discharged on 23-9-15 with following medication:

1. Syp. Phenytoin – 2.5ml, PO, BD.
2. Syp. Paracetamol – 3ml, PO, SOS.
3. Syp. Zincovit – 3ml, PO, OD.
4. Syp. Calcimax – 5ml, PO, TID.

Advise - To visit hospital after 2 weeks.

Discussion:

SWS is referred to as complete when both CNS and facial angiomas are present and incomplete when only one area is affected without the other. The Roach scale is used for classification -

Type I- Both facial and leptomeningeal angiomas; may have glaucoma

Type II – Facial angiomas alone; may have glaucoma

Type III – Isolated leptomeningeal angiomas; usually no glaucoma [7-9].

Only 8-20% of patients with facial port-wine birthmarks, with and without ocular involvement, develop neurological symptoms. Those with only V2 and/or V3 involvement have a significantly lower risk for experiencing symptoms of SWS, and early screening is less urgent [10,11]. 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 [12]. In our case birthmark was noticed on the right side of nose and is associated with neurological symptom of seizure, and hypocalcaemia with no ocular involvement. Her MRI report shows no abnormality. Brain - MRI with contrast, however, be advised that due to the progressive nature of SWS, brain involvement may not be evident on early scans. Early in infancy the abnormal blood vessels on the surface of the brain may not be seen even by MRI. If the initial MRI is normal, it can be repeated at 2-3 years of life to exclude the presence of a leptomeningeal angioma [13].

Treatment and prognosis depends upon the nature and severity of clinical features. So initially child was treated for seizures with anti-epileptic drugs whereas for port wine stain laser treatment was advised. Presently there is yet any specific treatment for SWS, and the management of the clinical manifestations and complications is still far from adequate [14]. Children with SWS often develop progressive problems including glaucoma, seizures, stroke, and intellectual disability, so a regular ophthalmological and neurological surveillance is needed till adulthood.

Conclusion:

Management of a patient with SWS may be challenging. The parents of the diagnosed patient must receive counselling concerning the potential risk of affected offspring. The health professionals have to be suitably able to recognize its characteristic signs and symptoms, and so improve the quality of life of the child. As this patient has neurological complications at an early stage of life chances of developmental delay are more and so there is a need for special education.

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