

## Case Report

### Sturge-Weber Syndrome

Hossain MA<sup>1</sup>, Karim AKMB<sup>2</sup>, Alam AMS<sup>3</sup>

#### Abstract

**Introduction:** *Sturge-Weber syndrome sometimes referred to as encephalotrigeminal angiomas, vascular malformation with capillary venous angiomas involving face, choroid of eye, leptomeninges, is a rare congenital neurological and skin disorder.*

**Case presentation:** *This is case report of 7 years old mentally disabled boy, with long-standing seizures, with a port-wine nevi on the right side of the face along the distribution of trigeminal nerve. Interictal encephalogram showed bilateral slow activity, pronounced in the right hemisphere, with epileptogenic activity in the right fronto-parietal region. Computerized tomography and magnetic resonance imaging showed intracranial calcifications, abnormally large veins in deep medullary and subependymal periventricular region, atrophy of the right hemisphere of brain and ipsilateral thickening of skull.*

**Conclusion:** *Professional counselling and support in addition to drug treatment can provide help to patients and their family to overcome their problems and improve the treatment outcome.*

**Keywords-** *Encephalotrigeminal angiomas, Vascular malformation, Port-wine nevi.*

#### Introduction

The Sturge-Weber syndrome is a neurocutaneous congenital but not an inherited disease and it occurs sporadically. It is a disorder of vasculature which belongs to the group of phakomatoses characterized by nevus flammeus and angiomas of the meninges<sup>1</sup>. It is a rare disease characterized by a birth mark called port wine nevi, associated with abnormalities of the brain caused by abnormal blood vessels (angiomas) that occur on the cerebral cortex<sup>2,3</sup>. These changes are usually unilateral. It can be seen in both sexes equally, and no racial differences have been identified. Port wine nevi are congenital malformations in the dermis of the skin involving venules, capillaries and possibly perivenular nerves<sup>4</sup>.

#### Case presentation

Mr. Jubair 7 years old mentally disabled boy living in a

rural part of Sirajganj, Bangladesh, developed seizures and left sided weakness since birth. At that time he was hospitalized and treated in local hospital. At the age of 2 years he has attended in OPD, Department of Neurosurgery, Khwaja Yunus Ali Medical College Hospital for control of seizure. The diagnosis, Sturge Weber syndrome is given to him based on clinical signs and radiological findings. The patient has received Phenobarbital 30 mg per day and used the medication for some months. Now he is on carbamazepine CR. Physical examination revealed port wine nevi, localized in the right site of the face, (Figure 1) with left sided hemi paresis. Phenobarbital was excluded by his mother, thus we started treatment with carbamazepine. In addition to the treatment of the patient, efforts were made to educate the family members about the regular treatment of the patient. Since then, patient has been seizure free for almost five years. The dose of

1. Dr. Md. Alamgir Hossain, Associate Professor, Dept. of Radiology & Imaging, KYAMCH, Enayetpur, Sirajgonj.

2. Dr. AKM Bazlul Karim, Associate Professor, Dept. of Neurosurgery, KYAMCH, Enayetpur, Sirajgonj.

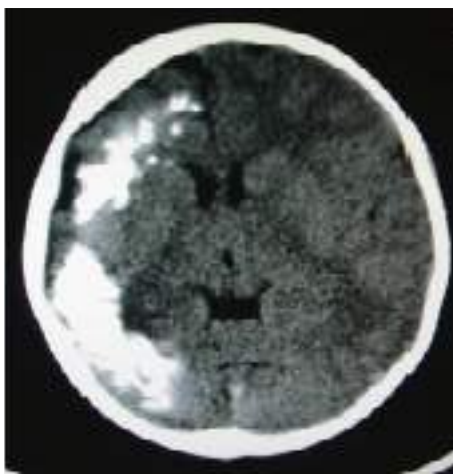
3. Dr. Abu Md. Shakurul Alam, Registrar, Dept. of Neurosurgery, KYAMCH, Enayetpur, Sirajgonj, Bangladesh.

carbamazepine is adjusted according to age and weight.



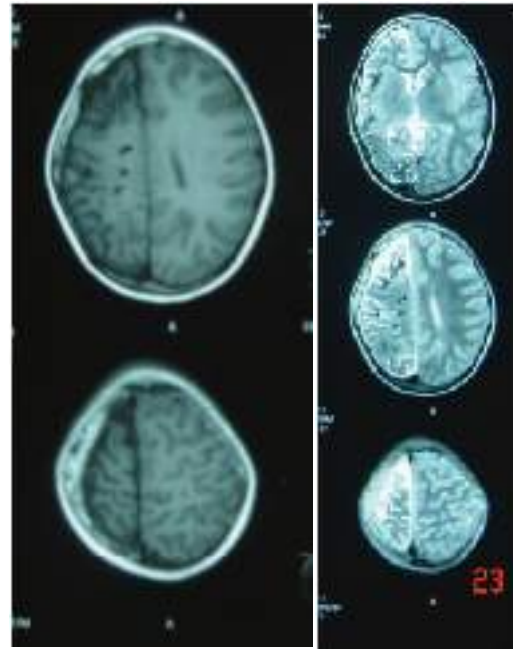
**Fig.1:**

Computerized tomography (CT) has been performed and the typical features of S.W. syndrome was revealed and showed gyriform calcifications with atrophy of right hemisphere and ipsilateral thickening of skull (Figure 2).



**Fig.2**

MRI of the brain was performed and it revealed severe right cerebral hemiatrophy, abnormally large veins in deep medullary and subependymal periventricular region. Left ventricle was wider compared to the right ventricle (Figure 3).



**Fig. 3:**

Interictal encephalogram showed bilateral slow wave activity, greater over the right side, with epileptogenic activity in the right fronto-parietal region. Neuropsychological examination was done and found- Psychological examination revealed IQ = 52 (Goodinaph test, Kohs test). He also showed latent aggressive tendencies and emotional imbalance.

**Discussion**

Sturge-Weber Syndrome is a congenital but not inherited disease. It is neurocutaneous syndrome presented with vascular malformations resulting from the failure of fetal veins to develop normally, changes in the brain, skin, and eye. These malformations lead to venous hypertension and subsequent hypoperfusion of the underlying cortex causing chronic cerebral ischemia, atrophy, and neurological deterioration. Sturge-Weber syndrome is a rare disease in the group of phakomatoses that cause physical, psycho-logical, and social disorders. This syndrome occurs with equal frequency in both sexes, with seizures typically developing in the first year of life<sup>5,6</sup>. This is a case report of a young patient who has type one of Sturge-Weber Syndrome according to the Roach Scale classification. It consists of cerebral calcifications, birth mark, seizures, glaucoma, hemiparesis, mental retardation, and cerebral atrophy<sup>7-9</sup>. Neurological deficit is caused by the intracranial vessels malformation<sup>10</sup>. Imaging findings consist of cortical calcifications - tram line calcifications, cortical atrophy,

enlarged ipsilateral choroid plexus, pial angiomas<sup>11</sup>. The best imaging modality is MRI while calcifications can be assessed in detail on CT images. The early onset of seizures prior to the age of 2 years is related to a poor prognosis with mental retardation, refractory epilepsy<sup>12</sup>, because of the larger involvement of brain dysfunction. Most cases with Sturge-Weber syndrome are not life threatening. This is a progressive disease, associated with continuous neurological decline<sup>13</sup>. With vigorous control and treatment of symptoms, such as seizures, visual problems, paralysis and mental disorder, quality of life can be preserved.

A detailed history, physical and mental state examination, neuropsychological, neuroimaging and laboratory investigations were undertaken in our case. Our patient developed seizures from birth. Sturge Weber syndrome has been reported in neonates as well - a case of 2 days old baby<sup>14</sup> and seizures are seen in about 75 to 90% of patients with Sturge-Weber syndrome<sup>15</sup>. Our patient had port - wine nevi on the right side of the face along the distribution of all three branches of the trigeminal nerve. Facial capillary vascular malformation-port-wine stain is common in the pediatric population, a study of 106 patients with port-wine stains<sup>16</sup>. Computerized tomography has shown the gyriphorm calcifications with atrophy of right hemisphere confirmed also with MRI associated with mental retardation. Cortical calcifications present at birth are reported in 30%<sup>17</sup>. In cases of development delay and mental retardation, 50 to 60% of patients with Sturge - Weber syndrome are affected<sup>15</sup>. In this case, seizures were controlled with Carbamazepine.

## Conclusion

In this case report we emphasize that regular treatment of the patient with Sturge-Weber with carbamazepine results in long term seizure free. A successful early treatment results in control of seizures and prevention of complications. Additionally, we strongly emphasize that professional counseling and support in addition to drug treatment can provide proper assistance to patients and their family to overcome their problems and improve the outcome of the treatment.

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