Sturge - Weber Syndrome

Mohammed Haneef

Koyili Hospital, Kannur, Kerala.*

ABSTRACT

Published on 30th December 2013

Sturge-Weber syndrome is a neurocutaneous syndrome caused by persistence of transitory primordial arteriovenous connection of the fetal intracranial vasculature. Diagnosis is considered when a child presents with seizure and facial capillary malformation along the trigeminal nerve distribution. Manifestations usually occur during early childhood. This case report describes a clinical presentation seen in adult life.

Keywords: Sturge Weber syndrome, Seizures, Facial malformation, Hamartoma, Phakomatoses.

*See End Note for complete author details

INTRODUCTION

Sturge-Weber syndrome is a neurocutaneous syndrome caused by persistence of transitory primordial arterovenous connection of the fetal intracranial vasculature. Diagnosis is considered when a child presents with seizure and facial capillary malformation along the trigeminal nerve distribution. Manifestations usually occur during early childhood. This case report describes presentation in adult life.

CASE REPORT

Thirty eight year old woman presented with first episode of focal seizures involving right side of the face and right upper limb for two days duration with out loss of consciousness. She is a diabetic and hypertensive on treatment for the past two years. Her facial appearance has been changing for the past ten years due to thickening of lips and increasing size of the nevus on the left side of the face. She had lost vision on her left eye a year ago, when she was diagnosed to have glaucoma. She is married and last child birth was sixteen years ago. On examination her blood pressure was 150/100, port wine stain involving left forearm (figure 1) and angiomas involving the lips were noticed.

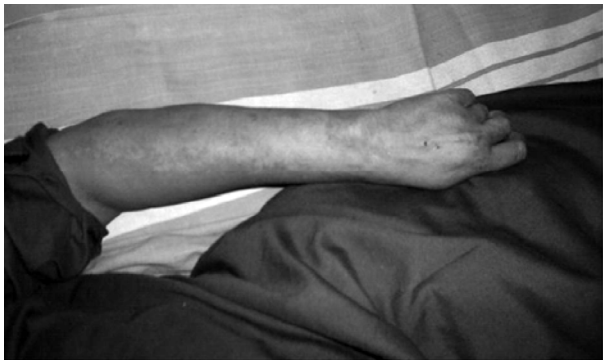

Figure 1. **Showing port wine stain left upper limb**

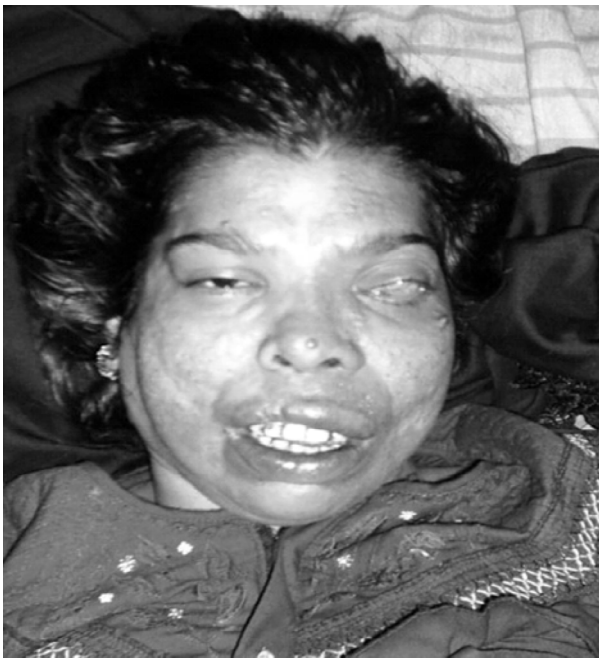

Figure 2. **Angioma lips and neovascularisation of left cornea**

Her left cornea was stony white with few blood vessels across its centre (figure 2).

Her neurological examination revealed focal seizures involving ® upper limb and ® side of the face in status. There was no focal neurological deficit and deep tendon reflexes were brisk on the ® side with an extensor plantar response on the same side. However

Corresponding Author:

Dr. Mohammed Haneef, MD (General Medicine), PGDHS (Diabetology), Consultant Physician and Diabetologist, Koyili Hospital, Kannur, Kerala. Phone: 9447236625. E-mail: dr.mhaneef@gmail.com

her other systems were within normal limits. Routine blood investigations were unremarkable. Her CT brain showed cortical calcification on left occipital region with cortical angioma and focal atrophy beneath the angioma (figure 3).

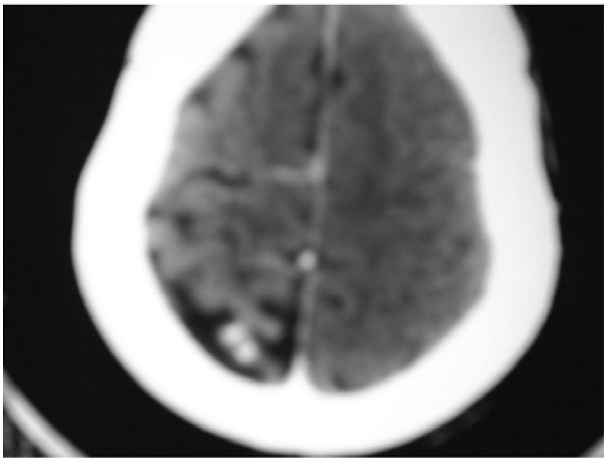

Figure 3. **CT brain- left occipital angioma and calcification**

She was treated with appropriate antiepileptics, antihypertensive and antidiabetic drugs. Seizures were controlled and she was discharged asymptomatic. Based on clinical features and CT findings a diagnosis of Sturge-Weber syndrome, epileptia partialis continua was made.

DISCUSSION

Sturge-Weber syndrome also known as encephalofacial angiomatosis belongs to a group of disorders collectively known as the phakomatoses. First description made by W.Allen Sturge in 1872 was in a six year child and the syndrome consists of congenital hamartomatous malformations that may affect the eye, skin, and central nervous system at different times.[1]

Three different types of Sturge-Weber syndrome are described:[2]

- Type I – most common type, includes port wine stain and brain angiomas
- Type II – port wine stain but no brain angioma
- Type III – brain angioma but no port wine stain

Clinically related features include focal or generalized motor seizures in as many as 85% of patients, some degree of mental retardation in approximately 60% of patients, and neurological deficits as hemiplegia. Focal or generalized motor seizures usually begin in the first year of life, and profound seizure activity sometimes may be observed with resultant further neurological and develop mental deterioration. Therefore, it is desirable to diagnose and treat the disease early, before permanent damage to the brain occurs.[1]

Calcifications in the external layers of the cerebral cortex underlying the angiomatosis associated with ipsilateral cortical atrophy frequently develop and progress with age, occasionally extending anteriorly to the frontal and temporal lobes.

The three forms of the syndrome generally are diagnosed on clinical grounds by the association of the typical cutaneous, central nervous system, and ocular abnormalities.

When a typical facial vascular skin lesion is found in a newborn, it should alert the physician to perform a complete ophthalmologic and systemic assessment for the potentially serious associated disorders. Children with bisymptomatic or trisymptomatic SWS initially may seem neurologically normal and have no symptoms of glaucoma or other ocular manifestations; thus, in some instances, the diagnosis may not become clear for an extended period of time.

The facial cutaneous lesion usually is the first component of the syndrome to be observed, since it is visible at birth. It may be very pale at first. Although it does not increase in extent, it usually becomes darker with age. Although not a medically threatening condition, the cosmetic deformity that the port-wine stain imposes may carry a psychological impact.

The glaucoma is almost always unilateral and ipsilateral to the port-wine stain, although contralateral or bilateral glaucoma with unilateral cutaneous lesions have been reported. The occurrence of glaucoma has been noted, especially when the facial skin changes involve the upper and lower eyelids. Numerous mechanisms have been postulated to explain the pathogenesis of glaucoma in SWS. At present, the most accepted explanation for the elevated intraocular pressure is a combination of developmental angle anomalies, which have a dominant role in infantile onset glaucoma and elevated episcleral venous pressure, which is more important in later onset glaucoma.[3]

Treatment of Sturge-Weber syndrome focuses on the symptoms. If seizures occur, antiepileptic medications such as carbamazepine, phenytoin, or valproic acid are given. Medications are used to reduce and control glaucoma and headaches. Laser treatment can lighten or remove the port wine stain on the face. Multiple treatments may be needed.[4]

END NOTE

Author Information

Dr. Mohammed Haneef, MD (General Medicine),
PGDHS (Diabetology),
Consultant Physician and Diabetologist,
Koyili Hospital, Kannur, Kerala.

Conflict of Interest: None declared

Cite this article as: Mohammed Haneef. Sturge - Weber Syndrome. Kerala Medical Journal. 2013 Dec 30;6(4):106-108

REFERENCES

1. Monte A Del Monte
2. Mary Kugler, RN
3. Arif Wahab. Sturge Weber syndrome – a review: Bombay hospital Journal, vol 50 No 1 2008
4. Thomas Sohl KA, Vaslow HT. Sturge Weber Syndrome: natural history and prognosis. J Epilepsy 1990; 3 (supply):293