RESEARCH PAPER | Medical Science | Volume : 6 | Issue : 2 | FEBRUARY 2016 | ISSN - 2249-555X

STURGE-WEBER SYNDROME- A LITERATURE REVIEW

KEYWORDS: Seizures, Encephalofacial angiomatosis, Port-wine stain, Computed tomography, Magnetic resonance imaging

Dr. Sanjeev kumar pandey
Junior Resident, Department of Medicine, Jawaharlal Nehru Medical College, Bhagalpur, Bihar

Dr. Prakash kumar
Junior Resident, Department of Medicine, Jawaharlal Nehru Medical College, Bhagalpur, Bihar

Dr. Rajiv Sinha
Medical Officer, Department of Medicine, Jawaharlal Nehru Medical College,, Aryabhatt Knowledge University, Bhagalpur, Bihar

ABSTRACT *The Sturge–Weber syndrome, also known as encephalofacial angiomatosis, is a rare neurocutaneous disorder that occurs as a sporadic congenital condition. It is characterized by a port wine stain that affects the skin in the distribution of the ophthalmic branch of the trigeminal nerve and is associated with venouscapillary abnormalities of the leptomeninges and the eye. It may be suspected in a child presenting with seizure and a characteristic skin lesion along trigeminal nerve distribution. The chacteristic features on computed tomography and magnetic resonance imaging lead to the diagnosis. The goal of therapy is centred around controlling the seizures.*

INTRODUCTION:

Sturge-weber syndrome also called as meningo- or encephalofacial (encephalotrigeminal) angiomatosis is a rare congenital neurological and skin disorder is part of a group of disorders known as phakomatoses (congenital neuroectodermoses)[1]. It is a rare syndrome, with an incidence estimated at 1 case in 20,000-50,000 persons[2] . It consists of congenital hamartomatous malformations that may affect the eye, skin, and central nervous system (CNS).

This condition has been referred to as Sturge-Weber syndrome,as it was W. Allen Sturge who in 1879,described a child with sensorimotor seizures contralateral to a facial "port-wine mark" and Parkes Weber (1922,1929),who gave the first radiographic demonstration of the atrophy and calcification of the cerebral hemisphere ipsilateral to the skin lesion.

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 6 th week of intrauterine development but normally undergoes regression during 9 th week[2].An associated gene mutation has been identified with nucleotide transition in *GNAQ* on chromosome 9q21 .

Angiomas of leptomeninges are usually unilateral, located in parietal and occipital region. Angiomas consists of a mass of enlarged and tortuous cortical vessels, supplied by one or more large arteries, and drained by one or more large veins. The presence of angioma results in alteration of vascular dynamics causing precipitation of calcium deposits in cerebral cortex underlying the angioma. Krabbe (1932,1934) showed conclusively that the calcification lay not in the blood vessels (as Dimitri and many others had concluded),but in the second and third layers of the cortex. Mental retardation, Seizures, Hemiparesis or Hemiplegia may develop secondary to this and severity depends on extent of the lesion[3].

A portwine stain is a cutaneous capillary malformation that occurs in approximately 3 of every 1000 newborns[2,4] and usually involves the head and neck in the distribution of the ophthalmic branch of the trigeminal nerve . A child born with a portwine stain on the face has approximately a 6 percent chance of having the Sturge–Weber syndrome[4], and this risk increases to 26 percent when the portwine stain is located in the distribution of the ophthalmic branch.

DISCUSSION:

Clinical manifestations:

Sturge Weber syndrome is a neurocutaneous syndrome (phakomatosis) with a sporadic occurrence. It has no racial or sex predilection. The typical patient presents at birth with facial angiomas, however the reverse is not always true. In the incomplete form of Sturge Weber syndrome, CNS angiomas occur without cutaneous manifestation thus no suspicion of the syndrome arises until the onset of seizures.

Clinical features:

A vascular nevus(port-wine stain) is observed at birth to cover a large part of the face and cranium on one side in the territory of the ophthalmic division of the trigeminal nerve(20 percent).Concomitant involvement of the maxillary or maxillary and mandibular branch occurs in many patients(2-23 percent).In 10-30 percent of the cases the nevus is bilateral[5].Orbiatal tissue,especially the upper eyelid,is almost invariably involved;congenital buphthalmos may enlarge the eye before birth and glaucoma may develop later in that eye,causing blindness.

The most common clinical manifestation is with childhood seizures, present in 71-89% of cases[6].In the majority of patients,they develop before 2 years of age[7].

Developmental delay and mental retardation are almost always associated with seizures. Other neurological manifestations include headache or hemiparesis.

Diagnosis:

The diagnosis of Sturge Weber syndrome is based on im-

Volume : 6 | Issue : 2 | FEBRUARY 2016 | ISSN - 2249-555X

aging studies, although CSF analysis may reveal elevated protein due to micro haemorrhages.

Skull x-ray were historically useful and capable of identifying the gyriform calcification of the subcortical white matter although they no longer play a significant role in the diagnosis or management of this condition. The finding usually becomes evident between 2 and 7 year of age[6].

On CT scan, homolateral cortical atrophy may be seen,which may be associated with a compensatory hypertrophy of the frontal bone and frontal sinus.Cortical and intracranial calcifications seen on CT scan have a characteristic 'tram-track' appearance.

MRI is the current "gold standard " for diagnosis of disease which is reliable even in very young infants[8].

Magnetic resonance imaging without contrast may show cerebral atrophy both in T1- and T2-weighted sequences,diploic prominence,and enlarged choroid plexus.MRI after contrast permitted a better evaluation of the extent and patency of the leptomeningeal angiomatous malformation,size of choroid plexus and the parenchymal venous anomalies.

Functional cerebral studies using PET and SPECT have shown abnormalities of metabolism and perfusion in Sturge- Weber syndrome.[9]

Treatment:
Treatment is aimed at controlling seizures pharmacologically, but surgical treatment may be necessary in refractory cases.

Medical treatment with sodium valproate,carbamazepine,phenobarbitol, or phenytoin have all been tried [5].Vigabatrin may also be useful.Facial port-wine stain can be treated with pulsed Dye laser.

Prognosis:
Seizures play a major role in failure of mental development and deterioration of mental function, hence effective seizure control is omnipotent. Factors associated with poor prognosis are:

1. Early onset seizures.
2. Seizures refractory to medical treatment
3. Extensive leptomeningeal angioma
4. Extensive atrophy of underlying cerebral cortex
5. Development of hemiparesis and
6. Deterioration in cognitive functioning

Conclusion:
The diagnosis and management of patients with Sturge-Weber syndrome requires the combined skills of a physician, radiologist and a psychologist.

Fig 1:Port-wine stain involving fore head and both upper and lower eye lids

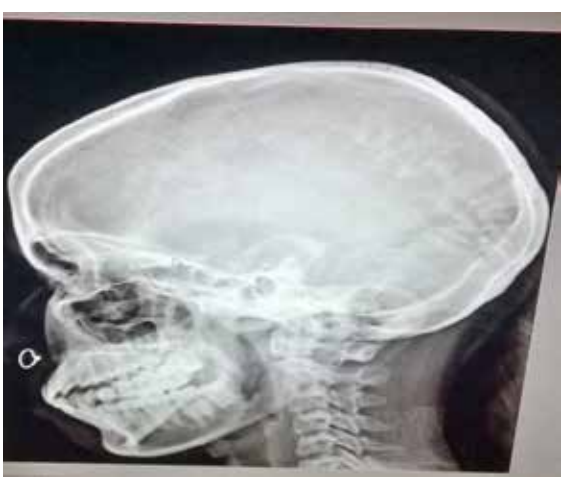

Fig 2: X ray of the skull in standard projections reveals intracranial calcifications in the form of "tram lines".

Fig 3: Cranial Computed Tomography shows cortical and sub cortical gyriform calcifications of the left brain hemisphere and cortical atrophy.

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