

CASE REPORT

STURGE WEBER SYNDROME-UNUSUAL PRESENTATION

N.S. Neki

Department of Medicine, Govt. Medical College and Guru Nanak Dev Hospital, Amritsar, Punjab, India

Sturge Weber Syndrome or encephalo-trigeminal angiomatosis is non-hereditary, congenital and rare disorder of unknown aetiology. It is characterised by vascular malformation with capillary venous angiomas involving face, eye and leptomeninges resulting in neurological and orbital manifestations. A case of 23 years old female presented with history of tonic-clonic convulsions, evidence of Port wine stain on face since birth, characteristic CT findings diagnosed as a case of Sturge Weber Syndrome is reported here for its rarity.

Keywords: Sturge Weber Syndrome; Port -wine stain; Seizures

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INTRODUCTION

Sturge Weber Syndrome (SWS) also called encephalo-trigeminal angiomatosis is congenital non-hereditary sporadic neurocutaneous disease. Shimer in the year 1860 described this syndrome for the first time and more specific description was given by Sturge in the year 1879 who described eye and skin changes as well as neurological manifestations. Weber in the year 1929 described radiological changes occurring in these patients.¹ Sturge Weber Syndrome is characterised by facial port-wine stain, ocular and neurological manifestations.^{2,3} The typical feature of this disease is the angiomas involving the leptomeninges, skin of the face especially the ophthalmic division of the trigeminal nerve. Neurological abnormalities include epilepsy, focal deficits, learning disorders, mental retardation including developmental delay, hemiplegia as well as orbital involvement resulting in buphthalmos and glaucoma.⁴ Dental manifestations may occur in the form of gingival hemangiomas lesion involving mandible, maxilla, lip, cheeks, palate, tongue, and floor of the mouth.⁵ A rare case of SWS presenting as seizures is described here.

CASE REPORT

A 23 year old female presented with history of generalized tonic-clonic convulsions for 2 days. There was no history of headache, vomiting, fever and tuberculosis. She had history of seizure disorder at the age of 18 years for which she was under medication. There was no history of delayed developmental stones and learning difficulties faced by the patient on questioning her parents. Her vitals were normal. On examination, she was conscious and well oriented. Clinical examination revealed erythematous hyper-pigmented plaque on the right face involving right side of upper lip also present since birth without encroaching left side of nose, lips and chin suggestive of superficial haemangioma. (Figure-1)

There were no signs of meningeal irritation. Examination of the oral cavity was unremarkable.

Laboratory investigations including hemogram, liver and kidney functions, blood sugar, electrolytes, and HIV test were normal. X-ray chest, X-ray skull, and ultrasound abdomen did not reveal any abnormality. Fundoscopy and cerebrospinal (CSF) examination were normal. Electroencephalogram (EEG) revealed focal and sharp waves confined to left parietal lobe. CT brain revealed hyperdense gyriform calcification in right parieto-occipital lobe (Figure-2). She was put on antiepileptic medication and became fully conscious on 5th day.



Figure-1: Erythematous hyper-pigmented plaque on the right face involving right side of upper lip

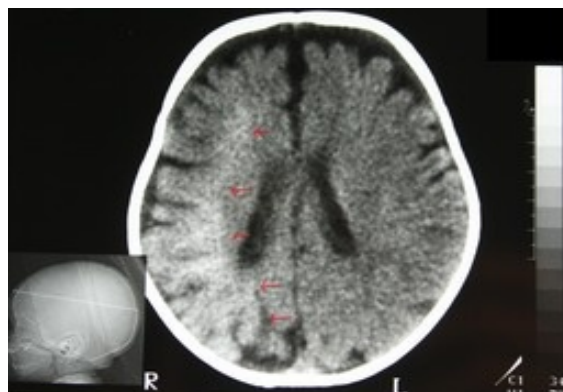


Figure-2: CT brain depicting prominent subcortical white matter calcification involving right parieto-occipital lobe

DISCUSSION

Sturge Weber Syndrome is characterised by port wine-stain over the face, ocular abnormalities (glaucoma and choroidal haemangioma) and leptomeningeal angiomas.² It is a rare disorder with incidence of 1:50,000 live births. Facial port-wine stain is a congenital macular lesion which initially may be light pink in colour, thereby involving area of distribution of trigeminal nerve as documented in this case. Neurological manifestations may vary from minimal or neurological signs to uncontrolled epileptic crisis, hemiparesis, hemiatrophy, mental retardation, microcephaly, and visual loss.⁶ They commonly involve right side of face especially the ophthalmic distribution of trigeminal nerve and usually do not extend beyond midline.⁷

Eighty percent of the patients with seizures show involvement of the contralateral side of Port-wine stain and the mechanism of the seizures is cortical irritation caused by the angioma resulting from hypoxia, ischemia and gliosis.⁷ Neuroimaging studies are of great help in the diagnosis, assessment of severity and progression of central nervous system (CNS) involvement in patients of Sturge Weber syndrome. X-ray skull especially lateral view may show tram like calcification which is usually in the parietal and occipital regions. Calcification is rare before 2 years of the age. CT brain reveals contrast enhancement of angioma, abnormal brain ependymal and medullary veins as well as abnormal enlarged choroid plexus ipsilateral to the angioma.⁸ Other modalities in the form of functional cerebral studies using Positron Emission Tomography (PET) and Single Photon Emission Computed Tomography (SPECT) reveal abnormalities of metabolism and perfusion in SWS.⁹ The treatment of SWS includes use of anticonvulsants, carbonic anhydrase inhibitors

for the control of glaucoma, beta-blockers for prevention of optic nerve atrophy and aspirin to prevent vascular disease and headache. As Port-wine stain presents cosmetic problems, it is treated with pulsed tunable dye laser. Surgical extirpation of affected lobe or hemispherectomy is indicated in resistant cases with rapidly progressive seizures in order to improve outcome with an aim to prevent developmental problems.⁹

CONCLUSION

Early diagnosis and management is recommended in view of varied clinical manifestations of Sturge Weber Syndrome with unknown exact aetiopathogenesis. The combined team efforts of physician, radiologist and psychologist are required for the better management of the SWS.

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Address for Correspondence:

Dr. N.S. Neki, Department of Medicine, Govt. Medical College and Guru Nanak Dev Hospital, Amritsar, Punjab, 143001, India.

Email: drneki123@gmail.com