A CASE REPORT OF STURGE WEBER SYNDROME
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Abstract: The Sturge-Weber Syndrome, also known as encephalotrigeminal angiomatosis, is a rare vascular neurocutaneous alteration. It is characterized by facial nevus, seizures, hemiparesis, intracranial calcification and mental retardation. Here we report a case of Sturge-Weber syndrome who presented with a cutaneous vascular nevus on the face during childhood as well as epileptic crisis episodes.

Keyword: sturge weber syndrome, encephalotrigeminal angiomatosis.

INTRODUCTION: Sturge-Weber syndrome belongs to a group of disorders collectively known as the phakomatoses (“mother-spot” diseases). It consists of congenital hamartomatous malformations that may affect the eye, skin, and central nervous system (CNS) at different times.

A CASE REPORT OF STURGE WEBER SYNDROME

A 30 year old male admitted with the history of left sided tonic clonic seizures lasting for 15 minutes associated with loss of consciousness. He had similar episodes of seizures which first started at the age of 3 years following which he was on treatment with antiepileptics. He was born out of non consanguineous marriage. He had developmental delay.

picture of the patient
Physical examination revealed pulse rate of 80 per minute, blood pressure of 130/80mmHg, height of 175cms, arm span of 165cms. There was port wine stain involving forehead extending up to the tip of the nose. He was conscious, oriented to time, place and person. Cranial nerve examination was normal. spinomotor and sensory system examination was normal.

Ophthalmoscopic examination revealed decreased visual acuity (6/60) with prominent episcleral veins in the right eye. Fundus examination was normal.

Psychological examination revealed IQ of 45%. Haematological and biochemical profile was within normal range. X-ray skull shows irregular calcification noted in the occipital region (tram-track sign). CT Brain showed irregular, tortuous, curvilinear, cortical calcification in the right occipital cortex region, no white matter edema. MRI Brain showed thickened cortex in the right occipito-parietal lobe with atrophic changes.T2W images shows hypointensity at cortical subcortical junction. T1 shows gyriform enhancement in right occipital lobe. MRA increased pial vascularity on right side. MRV shows engorgement of deep veins. He was treated with antiepileptics and supportive measures.

picture 3: arrow shows calcification

x ray skull lateral view- arrow shows calcification in occipital region T2W MRI- hypointensity in the subcortical region
This syndrome consists of constellation of symptoms and signs including a facial nevus, seizures, hemiparesis, intracranial calcification and mental retardation.

The hallmark of Sturge-Weber syndrome is a facial cutaneous venous dilation, also referred to as nevus flammeus or port-wine stain, which is present in as many as 96% of patients and is visible at birth. The facial venous dilation appears as one or several dull red patches of irregular outline, along, but not limited to, the distribution of 1 or more divisions of the trigeminal nerve. The neurologic manifestations vary, depending on the location of the angiomas, which most commonly are located in the parietal and occipital regions, and the secondary effects of the angioma. These include seizures, which may be intractable; focal deficits, such as hemiparesis and hemianopsia, both of which may be transient, called "strokelike episodes"; headaches; and developmental disorders, including developmental delay, learning disorders, and mental retardation. Developmental disorders are more common when angiomas are bilateral.

The 3 types of Sturge-Weber syndrome (THE ROACH SCALE)\(^6\) are as follows:
- Type 1 (Complete trisymptomatic Sturge-Weber syndrome) - All 3 organ systems are involved
- Type 2 (Incomplete bisymptomatic Sturge-Weber syndrome) - The involvement is either oculocutaneous or neurocutaneous
- Type 3 (Incomplete monosymptomatic Sturge-Weber syndrome) - Only neural or cutaneous involvement is noted. The primary defect is a developmental insult affecting precursors of tissues that originate in the promesencephalic and mesencephalic neural crest. These affected precursors then give rise to vascular and other tissue malformations in the meninges, eye, and dermis\(^8\). Neuroimaging can confirm the CNS involvement. Skull x-ray film may show classic "tram-line" or "tram-track" calcifications.

MRI has been reported to be superior to CT scanning in detecting the malformations affecting the CNS in Sturge-Weber syndrome\(^9\). MRI with gadolinium enhancement shows leptomeningeal angioma. It also demonstrates cerebral volume reduction and ipsilateral choroid plexus enlargement. In addition, intravenous contrast can demonstrate the curvilinear posterior contrast enhancement of ocular choroidal angiomas. On the other hand, CT scanning is superior to MRI in detecting the characteristic double-lined gyrfiform pattern of calcifications paralleling cerebral convolutions, referred to by radiologists as the railroad track sign as well as brain atrophy. However, these calcifications are usually not detectable before age 1 year and may not be seen for several years. SPECT demonstrates decreased cortical perfusion. PET demonstrates hypometabolism in areas that correspond to decreased perfusion\(^10\). EEG show electromagnetic changes in areas corresponding to the leptomeningeal angiomatosis. Treatment is symptomatic and focuses on seizure control with antiepileptics or surgery\(^11,12\). Symptomatic and prophylactic migraine management, glaucoma treatment to reduce intraocular pressure and laser therapy for facial cutaneous vascular malformation\(^13\). Physical therapy should be considered for infants and children with muscle weakness. Educational therapy is often prescribed for those with mental retardation or developmental delays, but there is no complete treatment for the delays.

DISCUSSION:
Sturge-Weber syndrome is a rare disorder that occurs with a frequency of 1: 50,000\(^7/4\). Both the sexes are equally affected and no racial difference has been reported. It is a sporadic neurocutaneous disease characterized by facial port-wine stain, ocular abnormalities (glaucoma and choroidal hemangiomata) and leptomeningeal angioma most often involving occipital and posterior parietal lobes.

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The prognosis of sturge weber syndrome is extremely variable. Bilateral brain involvement, early onset of seizures (less than 1 yr), refractory seizures predict more severe developmental and neurological problems.14

**BIBLIOGRAPHY**

4. Nelson textbook of paediatrics: chapter 589.3; mustafa sahin; p 2051-52
5. Choa DHL congenital neurocutaneous syndrome in childhood.III. sturge weber syndrome. *J .pediatrics* 1959;55;635
10. Chugani HT, mazzioleta jc,phelps ME, sturge weber syndrome:a study of cerebral glucose utilisation with PET *J pediatr* 1989:114:244
14. Rochkind s Hoffman HJ, Hendrich EB. sturge weber syndrome .natural history and prognosis *J Epilepsy* 1990,3(suppl):293