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Pediatrics

STURGE WEBER SYNDROME- A RARE CASE REPORT ORIGINAL

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ABSTRACT Sturge-Weber Syndrome (SWS) is one of the encephalotrigeminal angiomatosis and one of the important segmental vascular neurocutaneous disorders .The occurrence is not very uncommon and the prevalence is 1:20000 to 1:50000.(1) SWS occurs due to the presence of residual embryonic vessels . The various signs and symptoms include capillary malformation in the face a port wine birthmark and similar malformation in the brain involving leptomeniges as well as blood vessels of the eye causing glaucoma. The patient presents with seizures , hemiparesis and stroke like symptoms, headaches and developmental delay.(2) The imaging finding in SWS children is the calcification in the parietal and occipital area of the brain. The EEG findings in SWS are the attenuation and the excess of slow activities.We are presenting here a rare case of , a fourteen year old male child who presented to our emergency department with status epilepticus. The aim of presenting this case is to share the classical presentation and the challenges involved in the management.

KEYWORDS : Sturge-Weber Syndrome(SWS); leptomeningeal angioma: Port wine stain

CASE REPORT

My patient a fourteen year old male child presented to us with history of uncontrolled seizures in a state of status epilepticus. His past medical history revealed recurrent bouts of headache, most of the time severe in nature, along with right sided focal seizures with secondary generalization since 6 months of age. He was on long term antiepileptic treatment with multiple antiepileptic drugs (sodium valproate, clonazepam, clobazam and topiramate) for intractable seizures. He also had cognitive dysfunction with a poor scholastic performance. His IQ was 79. His anthropometric parameters were within normal limits. Hematological and biochemical profile was within normal range. The patient was communicative but apprehensive. He was born full term normal vaginal delivery with uneventful birth events. His family history was non contributory.

The general examination the patient revealed a unilateral portwine stain centered around right forehead, the right eye, side of the nose, right upper lip extending up to the midline. Her father revealed that the lesion was present since her childhood with progressive growth and caused slight discomfort when exposed to the sunlight.

Neurological examination of the patient in this child revealed subtle left hemiparesis, intractable seizures and the neuropsychological assessment revealed mildly challenged cognitive functioning.

Dental and oral examination of the patient showed a mixed dentition, with all teeth erupting at normal dental age and no other obvious hard tissue abnormalities.

Figs 1 &2- Port wine stain centered around right forehead, the right eye, side of the nose, right upper lip extending up to the midline

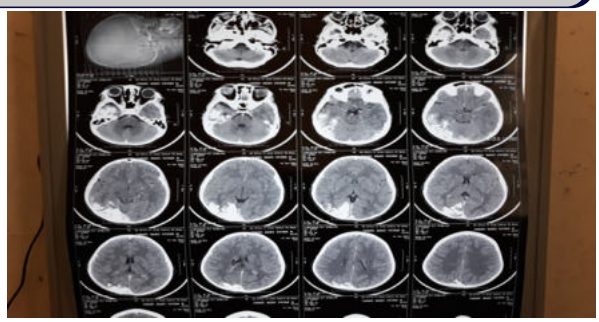

Fig- 3- Intracranial calcification s or Tramline gyriform calcification sign on CT brain

DISCUSSION

In 1879 Sturge described the association of a PWS with bupthalmos with focal seizures and postulated naevoid involvement of brain. The early radiological features of SWS were first reported by Weber 1922 and Dimitri 1923.(5)

Sturge Weber Syndrome is very rare disorder with a progressive lifelong morbidity. SWS may be complete when both CNS and facial angiomas are present & incomplete when only one is involved face or CNS. The commonest site is upper face & eyelids although lips, intra oral structures & pharynx may also be involved. Sometimes trunk and extremities of either side may be affected. Ipsilateral parietal and occipital regions are most commonly affected by the leptomeningeal angioma . Sometimes these findings could be bilateral. To begin with there is normal early development, however children with Sturge Weber Syndrome experience neurological and developmental deterioration in association with the onset of seizures and consequently suffer a high rate of disability. The classical imaging finding in these children is the evidence of calcification in the occipital area.(2) The striking features of the EEG in SWS are the attenuation and the excess of slow activities.(5)

The exact etiology remains controversial but sporadic incidence and focal nature of SWS suggests mutations. A single nucleotide variant in the GNAQ gene has been verified there is mosaic mutation in GNAQ gene. Low flow of the leptomeningeal capillary capillary malformation appears to result in chronic hypoxic state leading to cortical atrophy and cerebral calcifications.(1.2,3,4)

Clinical presentation in majority of patients includes seizures 75 -95% which are refractory and require several antiepileptic drugs. One third of patients develop hemiparesis on opposite side.50% of all SWS patients have mental retardation and some form of developmental delay&50% may get recurrent headache. Ocular problems include

glaucoma in 25% due to choroidal involvement, heterochromatic iris, nystagmus and the fundus may be of tomato ketchup color.(2,3) The evaluation includes contrast MRI cranium. By adolescence90% have typical linear parallel intracranial calcification typically described as tram track or rail track sign.

The management of these children is a challenge and involves a well coordinated multidisciplinary approach. This includes control of seizures and epilepsy surgery, corpus callosotomy and hemisherectomy in very resistant seizure cases. (6) Dermatological manifestations are managed by pulsed dye laser therapy and treatment should start during infancy itself. For the management of headaches it is symptomatic and prophylactic therapy only. Flunarizine has been found to be useful

CONCLUSIONS

To conclude, the approach to the management of these children requires a multidisciplinary approach involving primary care physician and pediatrician, health counselors, neurologists, Neurophysician as well as neurosurgeons, dermatologists, plastic surgeons & dental surgeons. The parents of all the diagnosed patients must receive counseling concerning the potential risk of the affected offspring.. The health professionals have to be trained . They should be able to recognize its characteristic signs and symptoms, and thus improve the quality of life of these SWS children.

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