

A INTRESTING CASE OF STRUGE WEBER SYNDROME

ABSTRACT:-

Struge Weber Syndrome is a rare neurocutaneous disorder characterized by Leptomeninigeal & facial angiomas mainly in the course of ophthalmic & maxillary branches of the trigeminal nerve. We report a case of 14 year old male who presented with left hemiparesis since 2 to 3 months & Generalized tonic clonic seizures since 5 days. Diagnosis is confirmed by CT/MRI. The classical findings seen on CT/MRI are atrophy and calcification of the cerebral hemisphere. The most common neurological symptom seen are seizures, they are typically focal tonic clonic seizures, seen during first year of life. They are often refractory to anti-convulsants. Most children present with slowly progressive hemiparesis during the course of illness.

Keywords: Struge Weber Syndrome, angiomas, seizures, CT/ MRI.

INTRODUCTION:-

Struge Weber Syndrome is a rare congenital neurocutaneous syndrome affecting the meninges & face. It was first described by Schirmer & later in 1879 more specifically by Struge, it is also known as leptomeningo facial angiomatosis & Struge Weber Dimitri Syndrome¹. In 1879 Struge described a child with sensory motor seizures contralateral to a facial "Port Wine Mark". Later Weber & Parkes who described the first radiographic demonstration of atrophy & calcification of the cerebral hemisphere ipsilateral to the skin lesion^[1]. The Facial Port wine stain is present at birth, it is unilateral, involving the upper face & eyelid in a distribution consistent with the ophthalmic division of the trigeminal nerve. The incidence of SWS has been reported to be 8-33% in those with portwine stain. The incidence of epilepsy in patients with SWS is 75-90%. They are typically focal tonic-clonic seizures, & they are present in most patients in the first year of life. The seizures are usually associated with a slowly progressive hemiparesis. Seizures are usually refractory to anticonvulsants^[5].

CASE REPORT:

A 14 year old, adolescent male presented with the chief complaints of progressive left sided weakness of the body since two to three months & generalized tonic clonic seizures since five days. Patient had the first episode of seizure at the age of four months, for which he was started on antiepileptics. The episodes continued as the treatment was irregular. Each episode lasted for about two to three minutes. It was associated with tongue bite and soiling of clothes, there was post-ictal loss of consciousness for five minutes. There was sudden onset weakness of left upper and lower limb three months ago. The weakness was progressive. There was no other neurological deficit. There is a history of developmental delay. CT scan of brain was done, which was suggestive of right cerebral hemisphere atrophy. All other lab investigations were normal.

General Physical Examination: Patient was conscious well oriented in time, place & person, pulse-78b/min, regular, all peripheral pulses were felt, BP: 110/70 mm of hg rt arm supine.

The gait was hemiplegic. power was 4/5 on left side at all joints. There was hypotonia on left side in upper & lower limb muscles. Psychological examination was suggestive of intellectual disability. Mini mental status examination – Score – 15, which was suggesting moderate cognitive impairment. Examination of face shows "PORT WINE STAIN" on the right side of face as shown in Figure No 1. all other systems were normal.



Figure 1. Portwine Stain on right side of the face

Figure 1. shows single large erythematous patch over right side of the face along the distribution of trigeminal nerve.

Eye examination was normal. Intraocular pressure was normal.

Oral examination revealed right sided gingival hypertrophy while the left side was normal. No signs of bleeding from any site.

Radiological Investigations :

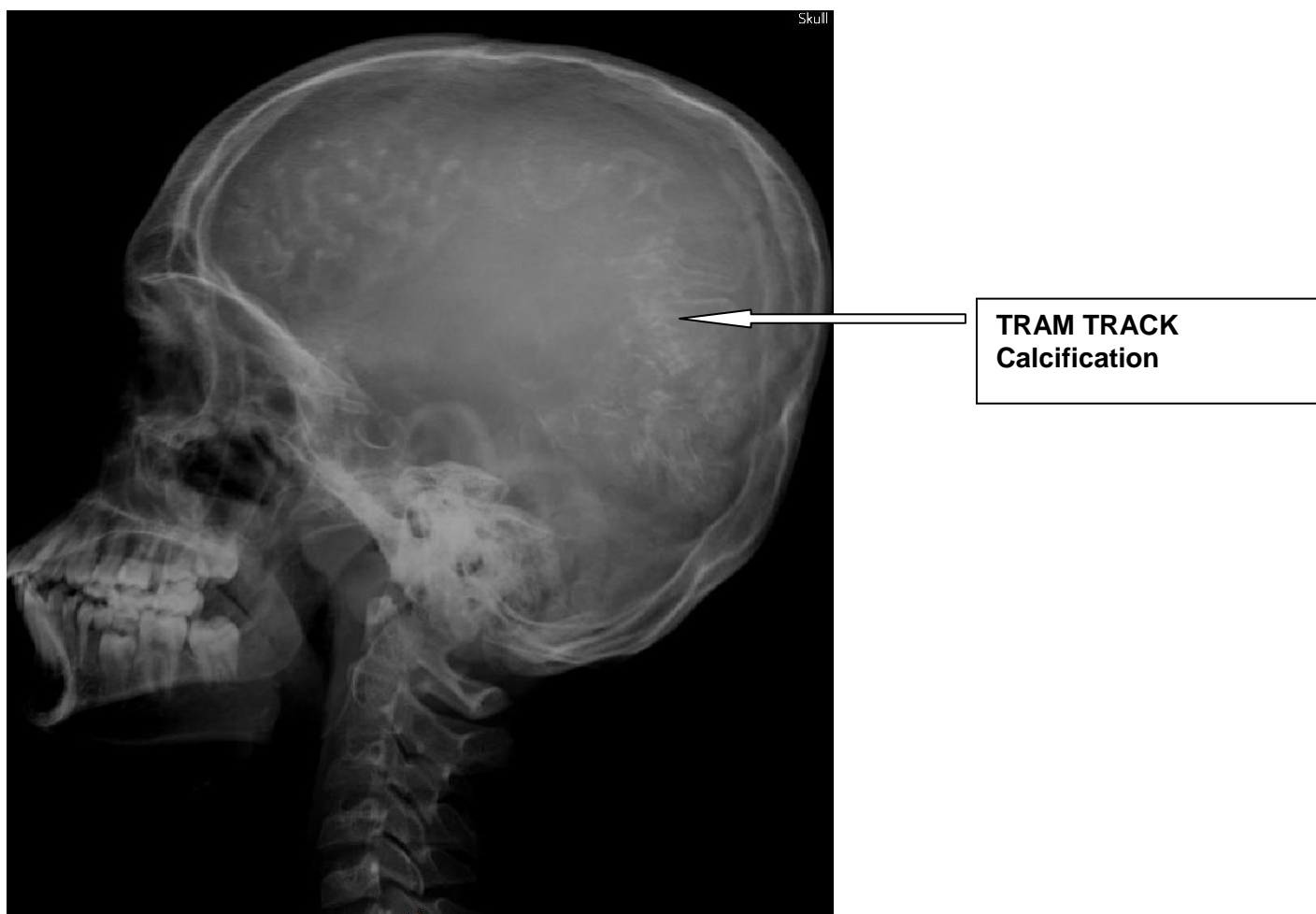


Figure 2. X-ray Skull – showing TRAM TRACK Calcification

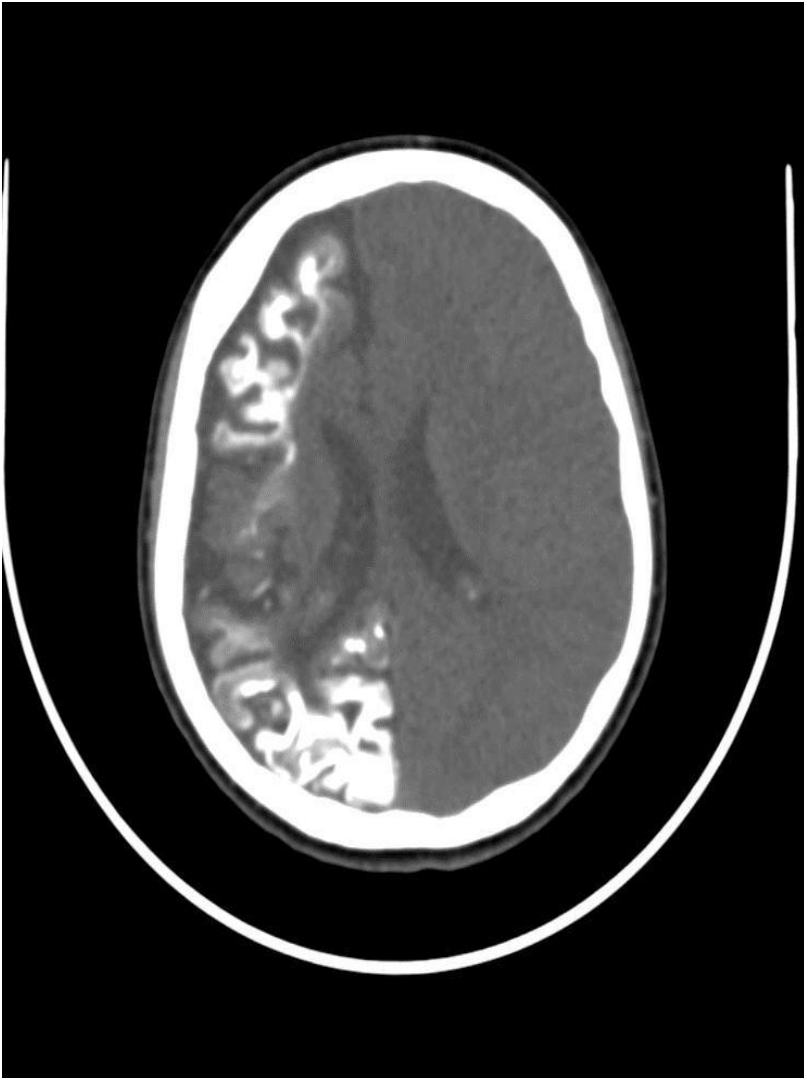


Figure 3. CT Scan Brain (Axial)

CT scan of the brain showed marked thinning of the gyri. There is prominence of the adjacent sulcal spaces on the right cerebral cortex. There is prominence of the lateral ventricle. Hyperdense gyriform calcifications and thickening of the calvarium are seen on the right side .Right cerebral hemisphere atrophy is also seen.

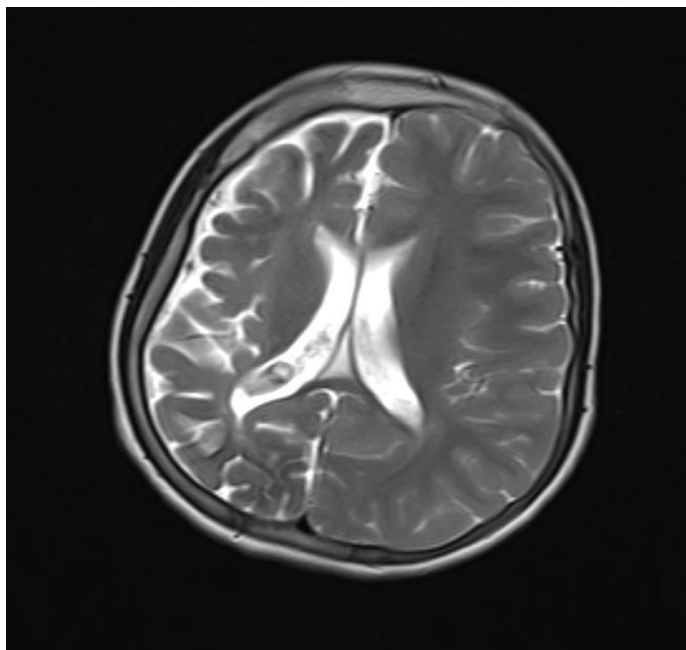


Figure 4. MRI Brain –T2WI

MRI Brain T2WI showed hyperintensities along the gyri on the right side.

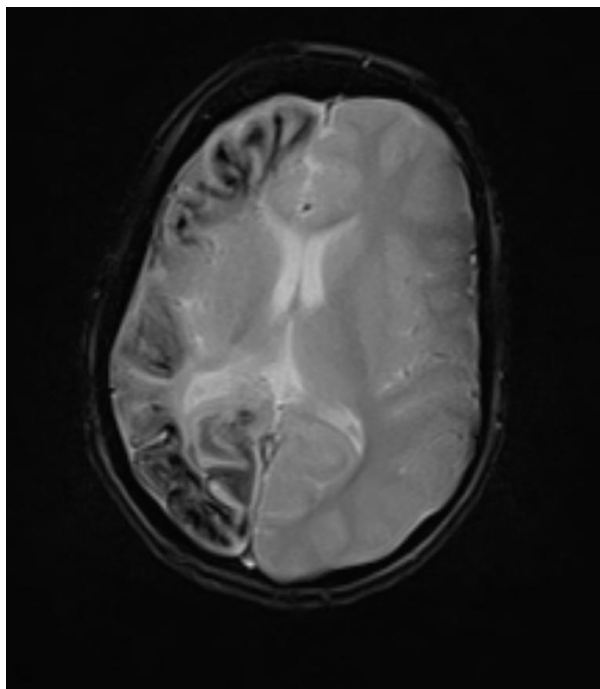


Figure 5. MRI Brain – GRE sequence

GRE sequence of MRI Brain showed blooming which appear hypointense . These are suggestive of calcifications.

EEG was performed which showed slow wave activity over the right side.

Based on the clinical history, physical examination and radiological evaluation , a diagnosis of Sturge Weber Syndrome was made.

Treatment:

Antiepileptics were started for the patient. Tablet Carbamazepine 100 mg two times a day was given. Patient's parents were asked to maintain the oral hygiene of the patient. Physiotherapy was advised for the weakness of the limbs. Professional counselling was also advised to overcome their social problems and to improve the outcome of the treatment. He was regularly being screened for further complications like bleeding. On his next visit after 3 months, the patient's relative confirmed that there were no episodes of seizure and that he is taking antiepileptics regularly.

DISCUSSION:

Sturge Weber syndrome or Encephalofacial angiomatosis belongs to a group of disorders known as phakomatosis[3]. SWS is a sporadic vascular disorder characterized by Facial Port Wine Stain [Facial Capillary malformation], Lepto meningeal Angiomas & abnormal blood vessels of the eye leading to Glaucoma[4,5]. Patients presents with intractable seizures, hemiparesis, stroke like episodes, developmental delay & mental retardation, most commonly involving the occipital & posterior parietal lobes.[3]. Incidence is 1 in 50000 persons[3,5].

It is a rare congenital but non-hereditary condition. The sporadic incidence & focal nature of SWS suggests the presence of somatic mutations. Whole genome sequencing from affected skin of some patients with SWS identified a single nucleotide variant [c.548G → A, P.Arg183Gln] in the GNAQ gene. This mutation has been confirmed in samples of affected tissue in larger cohort of SWS Patients, these results strongly suggests that SWS occurs as a result of mosaic mutations in GNAQ.[5].

It is caused by the presence of residual embryonal blood vessels and their secondary effects on surrounding tissues. There seems to be close relation between the persistence or maldevelopment of the embryonic vascular plexus of the eyelid & forehead & that of the occipitoparietal parts of the brain.[1]. The lesion in the cerebral cortex is replaced by glial tissue that calcifies, the explanation is that diversion of blood to the meninges during seizures causes progressive ischemia of the cerebral cortex. Seizures are responsible for the progressive neurologic deficits & efforts should be made to prevent them with aggressive anticonvulsant therapy[1].

Sturge weber syndrome is classified according to Roach's scale: -[5,6]

Type I – Both facial and leptomeningeal angiomas ; may have glaucoma.

Type II – Facial angiomas alone [no CNS involvement]; may have glaucoma.

Type III Isolated leptomeningeal angiomas ; usually no glaucoma

Neurologic manifestations vary and depend on location and extent of the leptomeningeal angioma. The most common neurological manifestations are seizures, spastic hemiparesis, some patients present with flaccid hemiparesis & wasting of muscles, hemisensory loss & homonymous hemianopia, all on the side contralateral to the trigeminal nevus. Skull Films & CT Scans taken second year after birth shows a characteristic

“Tramline” Calcifications involving the parietooccipital cortex [7]. CT/MRI shows Calcifications & underlying cortical atrophy. [1,2], MRI also shows pial enhancement on postgadolinium sequences, it is considered to be gold standard in assessing the extent of the disease. [1], it involves the occipital & posterior parietal cortex ipsilateral to the port wine stain. [1,8]. It has been emphasized that pluripotent neural crest cells within primitive embryologic segments known as metamers accounts for the distribution of cutaneous & neurovascular manifestations of several neurocutaneous syndromes including SWS [9].

Seizures are primarily sensory motor or generalized tonic clonic type or focal. [1,5]. Recurrent & repetitive seizure activity produces permeability changes in the blood brain barrier [10]. Several studies on SWS describes that cerebral-leptomeningeal enhancement increases transiently following seizure activity [11,12]. There is Refractory epilepsy not responding to anticonvulsants, surgical procedures including focal cortical resection, hemispherectomy and corpus callosotomy can be done but with less favorable outcomes...

Facial angiomas can be treated using dermabrasion, various pulsed-dye lasers, pulse light sources.

Conclusion:

Sturge weber syndrome is a rare neurocutaneous syndrome seen in children & younger adults. Presence of facial port wine stain can cause psychological trauma to the patient, & also affect his personality. Prompt diagnosis of SWS is essential for improving long term neurological outcomes. Demonstration of pial angioma is considered crucial for diagnosis of SWS, absence of pial enhancement on MRI findings should be interpreted with caution, particularly in presence of other neurologic manifestations of SWS. Children are at risk of developmental delay & intellectual disability. Treatment is symptomatic, it is aimed at controlling seizures, treating headaches & prevention of stroke like episodes. Glaucoma should be monitored with regular measurement of intraocular pressure. Our case of SWS is of Type I according to Roach's scale.

CONFLICT OF INTEREST: None, No Ethical issues.

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CONSENT: All appropriate consent forms taken.

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