Case study

STRUGE WEBER SYNDROME: A RARE CASE REPORT

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 3months & Generalized tonic clonic seizures since 5days.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 opthalmic 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[6].

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 seizures 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. Post-ictal loss of consciousness for approximately five minutes was also there.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 atropy.All other lab investigations were normal.

General Physical Examination:

The gait was hemiplegic .There was hypotonia on affected side.Patient was hemodynamically stable.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.

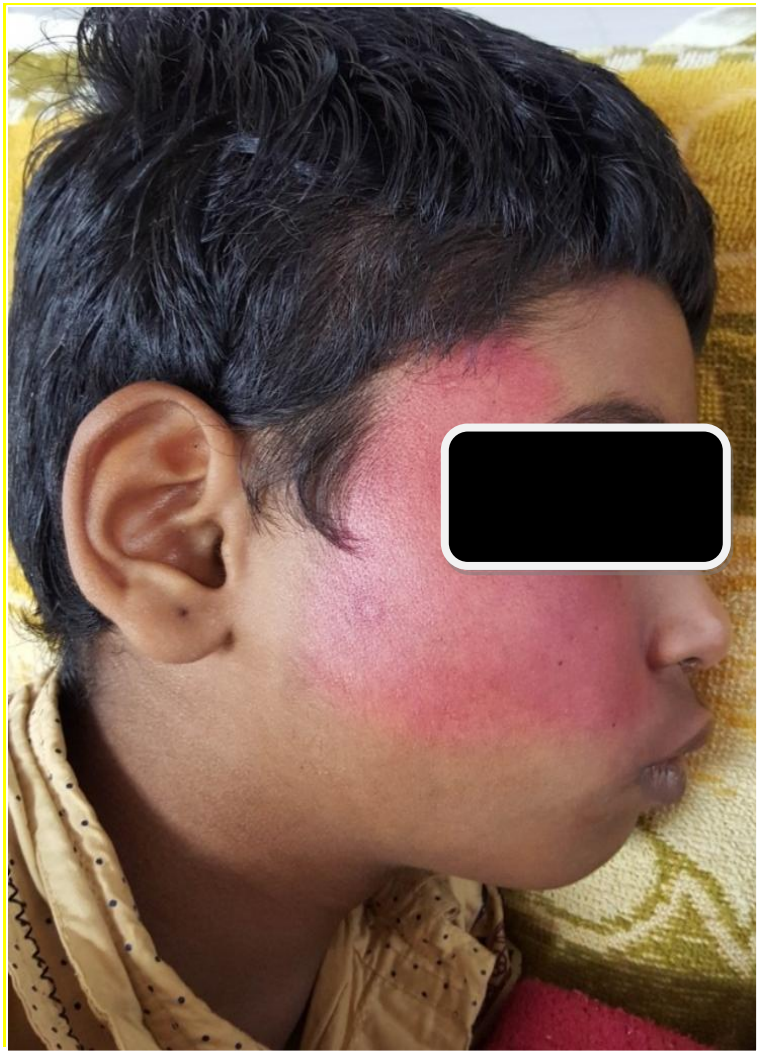

Figure 1. Portwine Stain on right side of the face

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 :

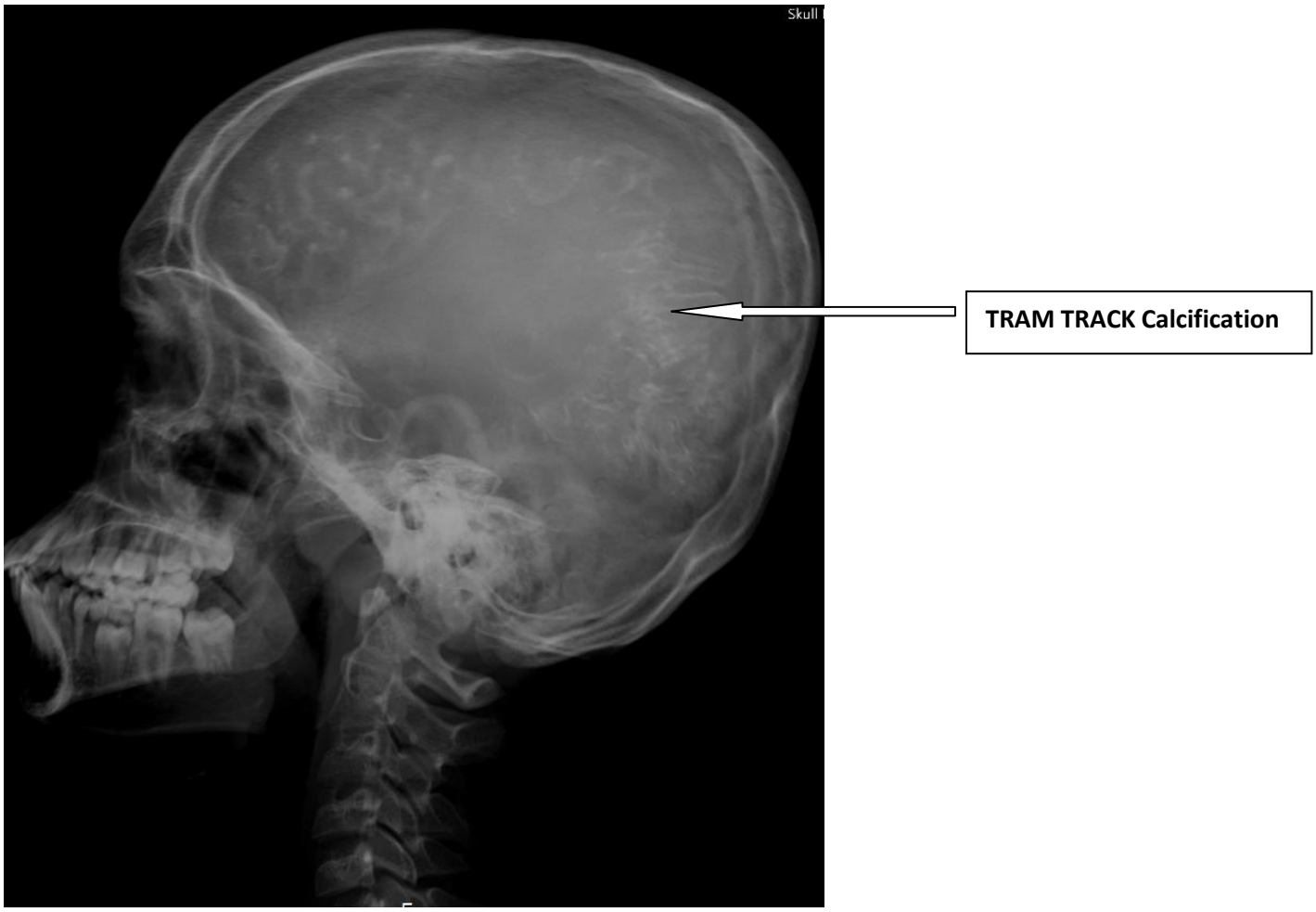

Figure 2. X-ray Skull – showing TRAM TRACK Calcification

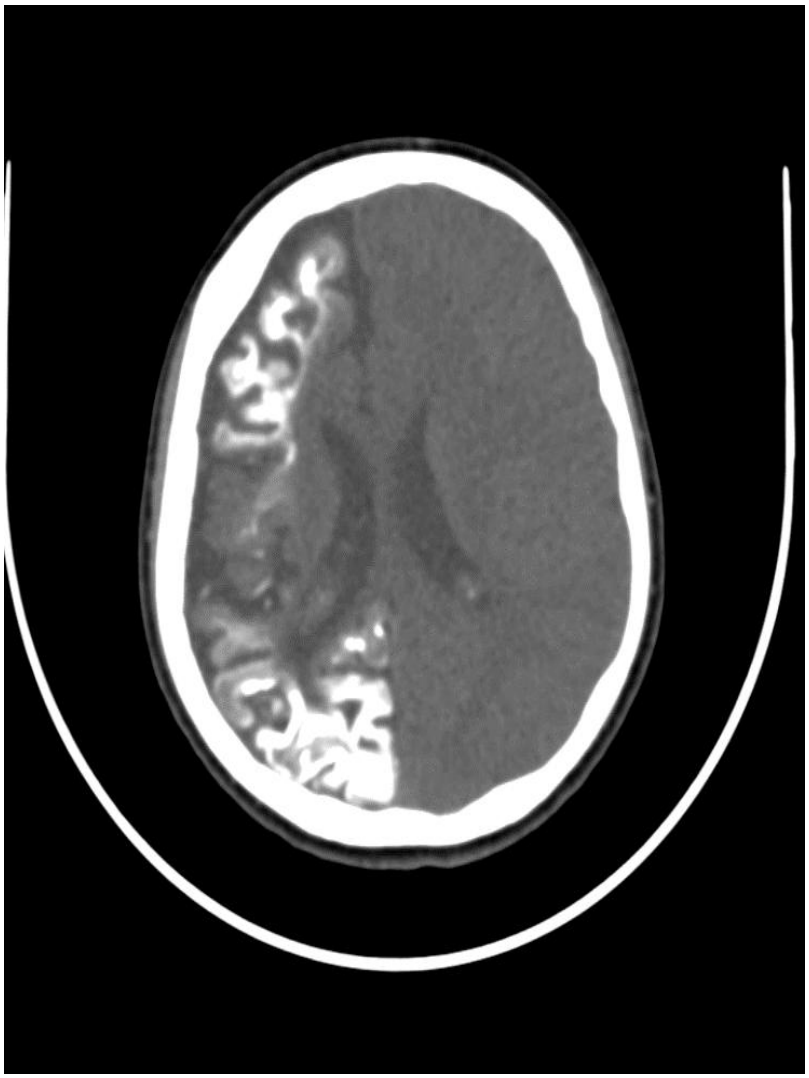

Figure 3. CT Scan Brain (Axial)

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 gyriformal calcifications and thickening of the calvarium are seen on the right side .Right cerebral hemisphere atropy is also seen.

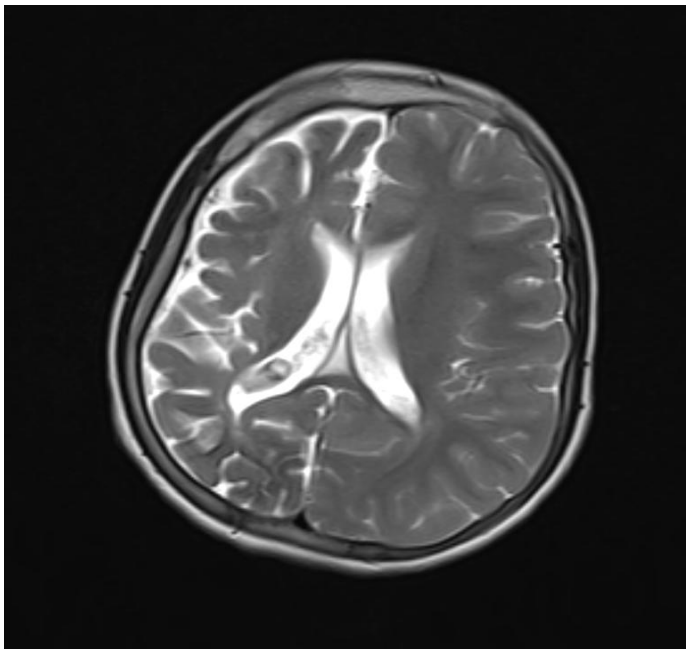

Figure 4. MRI Brain –T2WI

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

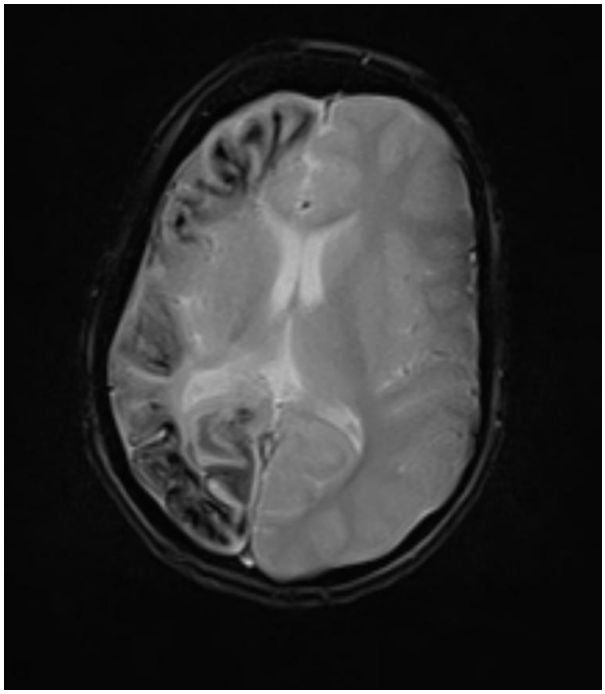

Figure 5. MRI Brain – GRE sequence

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[5,6].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[3,6].

It is a rare congenital but non-hered itary cond ition.The sporad ic incidence & focal natu re 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 stongly suggests that SWS occurs as a result of mosaic mutations in GNAQ.[6].

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 embryon ic vascular pleuxus 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 me made to prevent them with aggressive anitconulsant therapy[1].

Sturge weber syndrome is classified according to Roach's scale: -[6,7]

Type I – Both facial and leptomeningeal angiomas ; may have glaucoma.
Type II – Facial anngiomas 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 unilateral seizures,spastic hemiparesis,smallness of arms&legs,hemisensory loss& homonymous hemianopia, all on the side contralateral to the trigeminal nevus.Skull Films taken second year after birth shows a characterstic "Tramline"Calcifications involving the parietooccipital cortex.CT/MRI shows Calcifications& underlyling cortical atrophy.[1,2].
Seizures are primarily sensory motor or generalized tonic clonic type or focal.[1,6]. There is Refractory epilepsy not responding to anticonulsunts, surgical procedures including focal cortical resection , hemispherectomy and corpus callosotomy can be done but with less favoarable outcomes...
Facial angiomas can be treated using dermabrasion, various pulsed-dye lasers, pulse light sources.

Conclusion:

Struge weber syndrome is 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.Children are at risk of developmental delay & intellectuall disability.Treatment is symptomatic ,it is aimed at controlling seizures,treating headaches & prevention of stroke like episodes.Glaucoma should be mon itored with regular measurement of of intraocular pressure. Our case of SWS is of Type I according to Roach's scale.

REFERENCES:

1. Adams And Victor's –Principles of Neurology 11th Edition PG NO:1050-1051 ,Chapter 37 Developmental diseases of the nervous system..

2. Griffiths PD : Sturge Weber syndrome revisited : the role of neuroradiology.Neuropediatrics.1996,27:284-294.10.1055/s-2007-973796.

3. Thomas-Sohl KA,Vaslow DF,Maria BL. Sturge-Weber syndrome:a review.Pediatr Neurol.2004 May. 30 (5):303-10

4. Sharan S ,Swamy B, Taranath DA,et al.

5. Port-wine vascular malformations and glaucoma risk in Sturge-Weber syndrome . J AAPOS 2009 Aug.13(4):374-8

6 .Nelson Textbook of PEDIATRICS First South Asia Edition2016 ,Chapter 596.3 SWS by Musafa Sahin ,Page No:1879-1881

7 .Roach E.S,Neurocutaneous Syndromes ,Pediatric,clinical north am j.1992,39:591-620