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Original Research Paper

Radio-Diagnosis

STURGE WEBER SYNDROME: A CASE REPORT WITH CLINICAL AND RADIOLOGICAL FEATURES

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ABSTRACT Sturge–Weber syndrome, also referred to as encephalotrigeminal angiomatosis, is a rare congenital neurological and skin disorder. It is one of the phakomatoses and is often associated with port-wine stains of the face, glaucoma, seizures, mental retardation, and ipsilateral leptomeningeal angioma (cerebral malformations and tumors). It is characterized by abnormal blood vessels on the brain surface. We report a case of 8 year old male child with facial port wine stains along with radiological features which showed tram track sign and cortical atrophy in the right temporo-parieto-occipital lobe.

KEYWORDS : Sturge-Weber syndrome; encephalotrigeminal angiomatosis; clinical; diagnosis, differential

INTRODUCTION:

Sturge–Weber syndrome (SWS) is also known as encephalo-trigeminal angiomatosis. SWS was first described by Schirmer in1860. More specific description was given by Sturge in 1879. [1]

Sturge-Weber syndrome is a rare disorder that occurs with a frequency of approximately 1 per 40,000 to 50,000. Males and females are equally affected and there is no racial bias.[2] The disease is characterized by an intracranial vascular anomaly, leptomeningeal angiomatosis, most commonly involving parietooccipital region, followed by frontal and temporal lobes. Part or all of one hemisphere can be affected. SWS is unilateral in 80% cases and is typically ipsilateral to the facial angioma. Facial cutaneous vascular malformations, seizures, and glaucoma are among the most common symptoms and signs. Stasis results in ischemia underlying the leptomeningeal angiomatosis, leading to calcification and laminar cortical necrosis. Angiomas of leptomeninges are usually unilateral, located in parietal and occipital region. The presence of angioma causes alteration of vascular dynamics causing precipitation of calcium deposits in cerebral cortex underlying the angioma. Seizures, mental retardation, hemiplegia, or hemiparesis may develop secondary to this. Seizures are often medically refractory and worsen with time. Stroke-like episodes and focal neurological deficit is also seen.

The cutaneous angiomas are called port wine stains, that is plainly visible at birth. It can be unilateral (63%) or bilateral (31%) and is distributed over the skin innervated by the ophthalmic and maxillary division of trigeminal nerve. It may extend to chest, trunk and limbs.[4] Involvement of the area supplied by ophthalmic division is pathognomic.

Ocular involvement can result in diffuse choroidal hemangioma ("tomato catsup fungus), congenital glaucoma with an enlarged globe (bupthalmos), hemianopia and optic disc colobomas . [5] Intraoral angiomatosis may involve lips, buccal mucosa, palate, gingiva, and floor of mouth.

Case Report:

An 8 year old male patient reported to the department of radio-diagnosis, BSMC&H from paediatrics OPD for CT and MRI Brain with chief complaints recurrent seizures since birth, left sided upper limb and lower limb weakness and low intelligence. History revealed that the patient had perinatal asphyxia with hypoxic ischaemic encephalopathy-II. He has a younger sibling, born at full term by normal delivery. Family history was noncontributory.

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There were visible signs of mental retardation and patient had history of convulsions. Examination revealed port wine stain involving right side of face and trunk. [Fig.1&2]

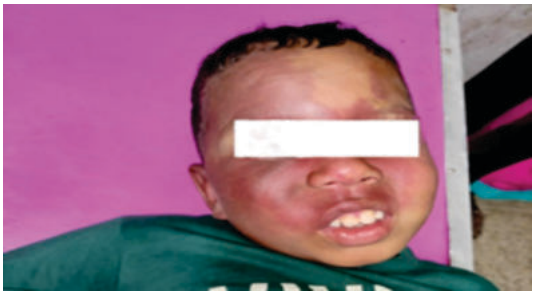

Fig.1 Portwine stain involving right side of face.

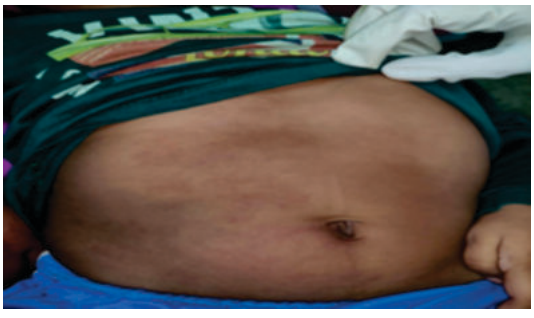

Fig. 2 Portwine stain on right side of trunk

Examination of eye had revealed bilateral sluggish pupillary reactions, and bilateral increased intraocular pressure (glaucoma).On radiological examination, X-ray showed the tram-track type calcification in the right parietal and occipital lobe with ipsilateral calvarial thickening [Fig.3]

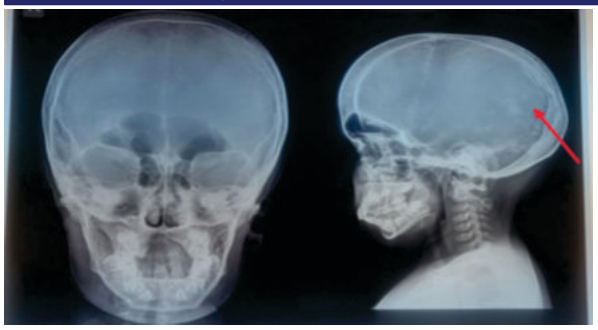

Fig. 3. AP & Lateral view X-ray of skull showing tram-track calcification in right parietal and occipital region with ipsilateral calvarial thickening.

NECT scan revealed atrophy involving right cerebral hemisphere with prominent sub-arachnoid spaces, predominantly involving the fronto-parietal lobes with evidence of tram-track or gyriform calcification pattern seen in right temporal- parieto- occipital lobe. [Fig. 4]

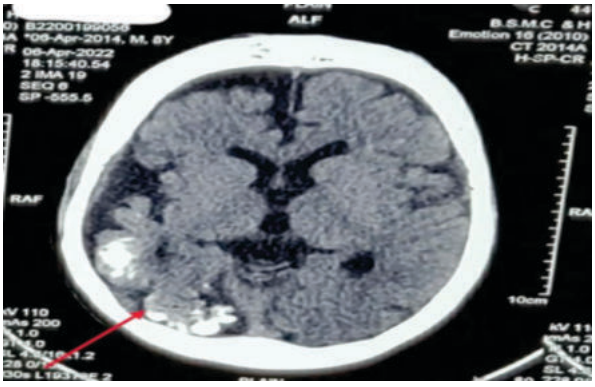

Fig. 4 NECT of Brain showing atrophy involving right cerebral hemisphere with prominent sub-arachnoid spaces, predominantly involving the fronto-parietal lobes with evidence of tram-track or gyriform calcification pattern seen in right temporal- parieto- occipital lobe.

MRI scan revealed atrophy involving right cerebral hemisphere with prominent choroid plexus in bilateral occipital horns and enlargement of adjacent sub-arachnoid spaces, predominantly involving the parieto-occipital lobes (on T1 and T2 & FLAIR sequences) [Fig.5,6&7] with evidence of tram-track or gyriform calcification (blooming on GRE sequence) [Fig.8] pattern seen in right posterior temporal, parietal and occipital lobes.There is calvarial thickening on right side.

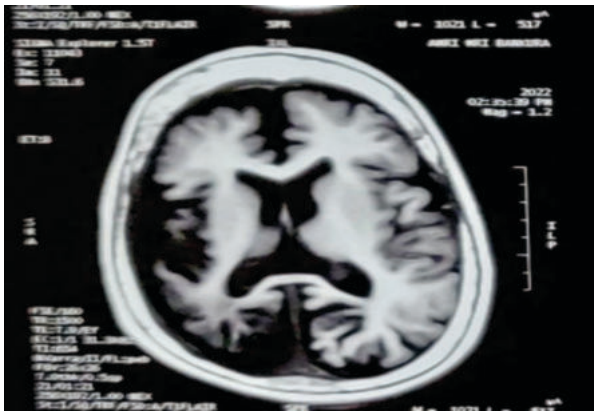

Fig.5 T1WI of Brain showing atrophy involving right cerebral hemisphere with prominent choroid plexus in bilateral occipital horns and enlargement of adjacent sub-arachnoid spaces, predominantly involving the parieto-occipital lobes

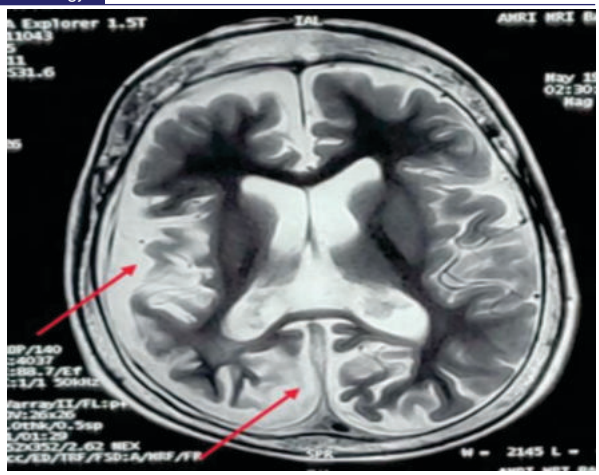

Fig. 6 T2WI of Brain showing atrophy involving right cerebral hemisphere with prominent choroid plexus in bilateral occipital horns and enlargement of adjacent sub-arachnoid spaces, predominantly involving the parieto-occipital lobes

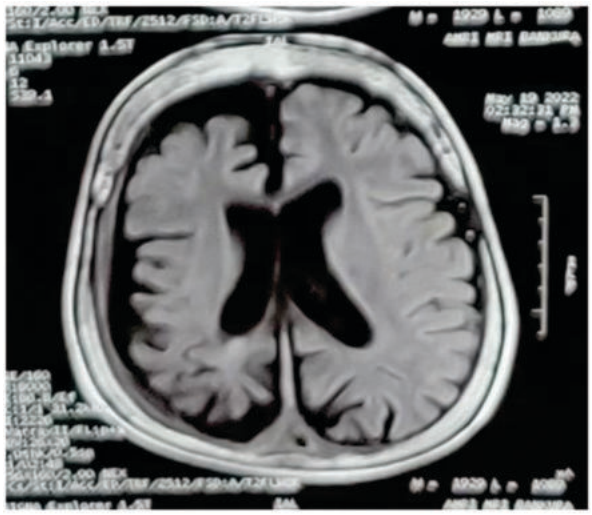

Fig. 7 FLAIR sequence of Brain showing atrophy involving right cerebral hemisphere with prominent choroid plexus in bilateral occipital horns and enlargement of adjacent sub-arachnoid spaces, predominantly involving the parieto-occipital lobes

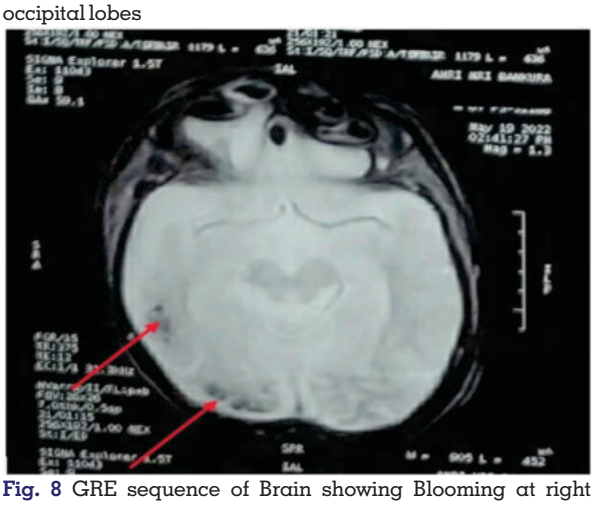

Fig. 8 GRE sequence of Brain showing Blooming at right parietal and occipital lobe.

With the help of clinical and radiological findings a diagnosis of sturge weber syndrome was made.

DISCUSSION:

Port wine stains represent hamartomatous capillary malformations and are named so due to the deep red hue that they leave on the skin or mucosa. [6] Such lesions characteristically bleed profusely when traumatized. In our case, the patient had port wine stain on his right side of face and trunk. Not all patients with facial port wine stains have Sturge – Weber angiomatosis.Only patients with involvement along the distribution of the ophthalmic branch of trigeminal nerve are at risk for the development of full condition. [3] SWS may also show oral manifestations. These include unilateral vascular hyperplasia of the oral mucosa and/or gingival changes ranging from slight vascular hyperplasia to large masses, which may interfere with mouth closure. [9]

SWS is referred to as complete when both CNS and facial angiomas are present and incomplete when only one area is affected without the other. The Roach scale [7] is used for classification:

Type I: Represents classic syndrome, Both facial and leptomeningeal angiomas; may have glaucoma.

Type II: Facial angiomas without evidence of intracranial disease; may have glaucoma.

Type III: Isolated leptomeningeal angiomas; usually no glaucoma.

Our patient had only facial angioma with glaucoma which comes under Type II of SWS according to Roach scale. Endocrine disorders are a newly recognised aspect of SWS. These patients have increased risk of growth hormone deficiency and central hypothyroidism.

Differential Diagnoses

Differential diagnoses are a combination of that for multiple intracranial calcification, cerebral hemiatrophy and leptomeningeal enhancement. Differential diagnosis includes (1) Blue rubber bleb nevus syndrome (2) Wyburn-Mason syndrome (3) Parkes- Weber syndrome- capillary malformations and limb overgrowth but also have arterio-venous fistulas in skin, muscle, bone, brain.(4) Hereditary hemorrhagic telangiectasia- condition characterized by abnormal dilatation of terminal vessels of skin, mucosa and occasionally viscera. (5) Angio-osteodystrophy syndrome-characterized by port wine stains on face, varices and hypertrophy of bones. (usually long bones) (6) Maffuci's syndrome-characterized by multiple angiomas of skin and chondromas of bone. (7) Von Hippel Lindau disease- a familial syndrome involving hemangioblastoma in retina and cerebellum. (8) Klippel Trenaumy-Weber syndrome is a syndrome characterized by combination of capillary malformation, soft tissues, bone hypertrophy and venous malformations[4, 8].

The diagnosis is based on clinical and imaging studies. Port wine nevus is observed clinically. Skull films may reveal tram track calcification caused by calcification in apposing gyri, ipsilateral calvarial thickening and enlargement of the paranasal sinuses and mastoid. Cranial NECT scan revealing cortical/subcortical tram-track calcifications in the brain underlying the angioma. In our patient, cranial NECT showed cortical atrophy with tram track calcification. Bone CT shows thickening of the diploe and enlargement with hyperpneumatization of the ipsilateral frontal sinuses secondary to long standing volume loss in underlying brain. MRI is the current gold standard for diagnosis of the disease which is reliable even in very young infants. ([10]) T1 and T2 scans show volume loss in the affected cortex with enlargement of adjacent subarachnoid spaces. Dystrophic cortical/subcortical calcifications are seen as linear hypointensities on T2 that bloom on GRE. FLAIR scans may demonstrate serpentine hyperintensities in the sulci, the "ivy" sign. DWI is usually negative. Post-contrast T1 and FLAIR sequences best demonstrate the pial angioma. Angiography demonstrates a lack of superficial cortical veins with corresponding dilatation of deep medullary and subependymal veins. MRI of our patient showed atrophy of right cerebral hemisphere and prominent subarachnoid spaces in fronto-parietal lobes with prominent choroid plexus in bilateral occipital horns.

Management:

New born babies with a port wine stains should have an ophthalmological examination in the first month of life, followed by neuroimaging (CT and gadolinium enhanced MRI) by 6-12 months age or sooner if neurologic signs are present. Cerebral blood flow imaging, Single Photon Emission Computed Tomography (SPECT) and Positron Emission Tomography (PET) are also useful when possible. The neurological signs are due to ipsilateral leptomeningeal angioma involving the occipital and posterior parietal lobes of the brain; vascular stasis with resultant ischemia leads to calcification and laminar cortical necrosis.([13])

Management of the syndrome involves both medical and surgical approaches. Medical treatment includes anticonvulsant therapy. Seizure control is achieved in less than half of the cases. Acute rescue treatment of seizures with benzodiazepines or if ineffective, intravenous phenytoin or phenobarbitone is recommended in India.([11])

PWS may be treated with cosmetic camouflage creams, pulsed tunable dye laser and cosmetic surgery. Laser therapy is the most effective approach to therapy of the PWS, but results are extremely variable being considered satisfactory in only 45% cases ([13]). Glaucoma is treated with timolol and lantaprost or surgical approaches to drain fluid from the eye to relieve excessive intra-ocular pressure, when medical therapy is unsuccesful. Low dose aspirin decreases the frequency and severity of stroke- like episodes and seizures.([14]) Early lobectomy or hemispherectomy in infants with drug-resistant epilepsy and widespread hemispheric angioma may be an option in severe cases. ([12])

REFERENCES:

1. Caiazzo A, Mehra P, Papagearge MB. The use of preoperative percutaneous transcatheter vascular occlusive therapy in the management of Sturge-Weber syndrome – Report of a case. J Oral Maxillofac Surg. 1998; 56: 775–8.
2. Zaroff CM, Isaacs K. Neurocutaneous syndromes: behavioral features. Epilepsy Behav 2005; 7: 133-142.
3. Neville BW, Damm DD, Allen CM, Bouquot JE, editors. 3rd ed. St. Louis: Elsevier; 2009. Oral and Maxillofacial Pathology.
4. Suprabha B, Baliga S. Total oral rehabilitation in a patient with port wine stains. J Indian Soc Pedod Prev Dent. 2005; 23: 99–102.
5. Gorlin RJ, Pindborg JJ. New York: McGraw- Hill; 1964. Syndromes of head and neck; pp. 406–9.
6. Fishman SJ, Muliken JB. Hemangiomas and vascular malformations of infancy and childhood. Pediatr Clin North Am. 1993; 40: 1177–200.
7. Roach ES. Neurocutaneous syndromes. Pediatr Clin North Am. 1992; 39: 591–620.
8. Mukhopadhyay S. Sturge –Weber syndrome: A Case report. J Indian Soc Pedod Prev Dent. 2008; 26: 29–31.
9. Yukna RA, Cassingham RJ, Carr RF. Periodontal manifestations and treatment in a case of Sturge –Weber syndrome. Oral Surg Oral Med Oral Pathol. 1979; 47: 408–15.
10. Comi AM. Sturge-Weber syndrome and epilepsy: an argument for aggressive seizure management in these patients. Expert Rev Neurother2007; 7: 951-956.
11. Rochkind S, Hoffman HJ, Hendrick EB. Sturge–Weber syndrome: natural history and prognosis. J Epilep1990; 3: 293.
12. Sarah A. Sturge Weber syndrome: A Review. Ann Indian Acad Neurol 2007; 10: 55-58.
13. Puneet Yadav, Shifa Yadav, Jai Chowdhary, Pawan Jain, Hemant Kumar Mishra. "Sturge Weber Syndrome: A Case Report with Clinical and Radiological Features". Journal of Evolution of Medical and Dental Sciences 2014; Vol. 3, Issue 46, September 22; Page:11305-11309, DOI:10.14260/jemds/2014/3476
14. Comi, Anne. "Current Therapeutic Options in Sturge-Weber Syndrome." Seminars in pediatric neurology vol. 22,4 (2015): 295-301.doi:10.1016/j.spen.2015.10.005