



Spectrum of Magnetic Resonance Imaging Findings in Sturge Weber Syndrome: A Rare Neurocutaneous Syndrome

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Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

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Case Report

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ABSTRACT

Sturge Weber syndrome is a type of neurocutaneous disorder also known as meningofacial angiomatosis. It is generally seen in paediatric age group and more commonly affects the males which present with delayed milestones, seizure disorder, loss of vision and other neurological deficits. Clinical suspicion of Sturge weber syndrome should be made on the presence of port wine stain (facial nevus) in a young children. MRI (magnetic resonance imaging) plays a key role in detecting the various spectrum of cortical and vascular defects associated with the syndrome. Clinicians can come to a final diagnosis of Sturge weber syndrome, when the clinical history is supplemented with MRI findings. We in our case report will be discussing the various spectrum of MR findings in this rare neurocutaneous syndrome.

Keywords: Sturge weber syndrome; MRI; pial angiomatosis; port wine stain.

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1. INTRODUCTION

Sturge Weber syndrome(SWS) is one of the rare paediatric condition, classified under neurocutaneous disorders having varying clinical presentations ranging from facial nevus (also known as port wine stain) to central nervous system manifestations like – mental retardation, seizure disorders , glaucoma, ocular choroidal haemangioma and hemiparesis [1].

The clinical presentation was originally described by Sturge in the year 1879 which included fascial port wine stain, glaucoma and focal seizures which was followed by radiological correlation by Weber in 1922 which suggested ipsilateral brain sclerosis (atrophy) as one of the underlying cause [2,3]. However facial port wine stain (PWS)is not synonymous with Sturge Weber syndrome, PWS can present as an isolated entity. Only in patients having PWS distributed along the ophthalmic division of trigeminal nerve involving the upper face and eyelid and underlying neurocutaneous syndrome like Sturge Weber should be suspected [4].

According to Roach et al. Sturge weber syndrome can be classified into 3 categories- **Type I:** representing classic syndrome, having both facial and leptomenigeal angiomas; may or may not have glaucoma, **Type II:** presents with facial angioma without evidence of intracranial disease; may or may not have glaucoma, **Type**

III: presents with isolated leptomenigeal angioma; usually no glaucoma [5]. Over the years various imaging findings on conventional radiographs, CT , MRI have been described, we in our case report will presenting a classical case of Type 1 Sturge weber syndrome and discussing the various neuroimaging findings.

2. CASE PRESENTATION

A 8 year old male child with a clinical diagnosis of Sturge weber syndrome presented to our radiology OPD having a history of mental retardation, delayed development, left sided hemiparesis and multiple GTCS (general tonic clonic seizures) since infancy. The patient currently complained of decreased vision, redness and purulent discharge from the left eye since last 10 days which had increased in intensity in the past 3 days. The patient was referred by the ophthalmology and internal medicine department for a MRI (magnetic resonance imaging) brain and orbit with contrast.

General physical examination revealed the presence of red pigmentation of the skin (port wine stain) predominantly involving the left half of face, crossing the midline and involving the medial aspect of right face. The left orbital region showed soft tissue oedema with ipsilateral exophthalmos and features of conjunctivitis (Image 1).



Image 1. Patchy areas of red discoloration of skin over upper left and right side face

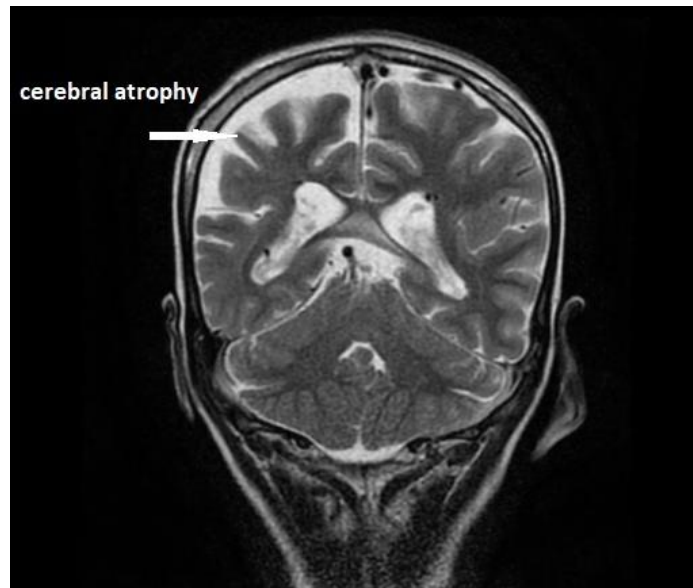


Image 2. Coronal T2WI shows prominent sulcogyral spaces in the right parietal region s/o cerebral atrophy (solid white arrow)

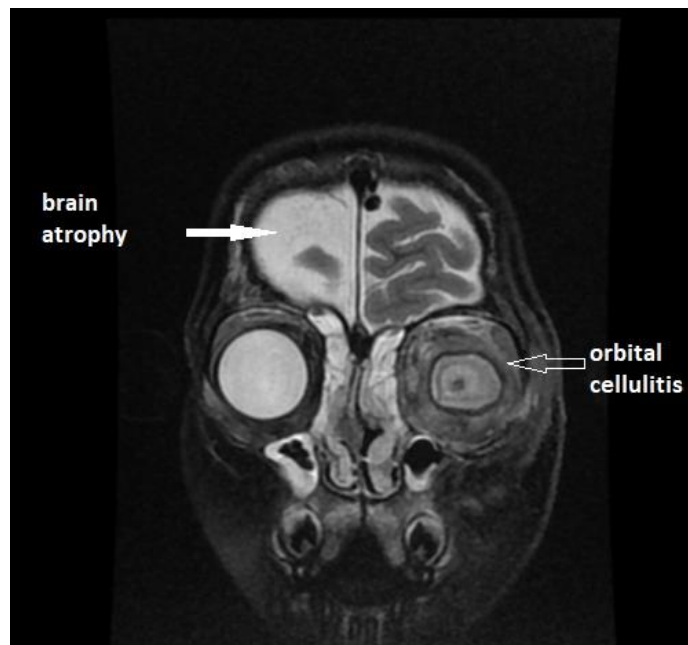


Image 3. T2WI coronal image showing loss of right sided frontal brain parenchyma (solid white arrow), intra conal and extraconal compartments of left orbit show oedematous fat and orbital muscles with altered signal intensity (white arrow)

MRI brain with orbit revealed- i) The presence of bilateral cerebral atrophy (right>left) with associated leptomenigeal enhancement in right parietal and bilateral occipital region. ii) Bilateral enlarged choroid plexus with vivid post contrast enhancement suggestive of choroidal angioma.

iii) Presence of few areas of altered signal intensity in the right parieto occipital region in the sub cortical white matter, appearing hypo intense on T1/T2WI and blooming on GRE which were suggestive of tram track calcifications. iv) The left orbit showed intense post contrast enhancement

in its intra and extra conal compartments, involving the ocular muscles, lense, anterior chamber, vitreous and the choroid , there was further thickening and enhancement of left optic nerve all these findings suggested pan ophthalmitis. v) In addition to the above

mentioned findings there was presence of increased deep medullary veins with involvement of cavernous sinus and thrombosis of the superior sagittal sinus, torcular herophili , distal straight sinus and right transverse sinus(Images 2-6).



Image 4. Axial contrast enhanced image depicts vivid contrast enhancement in the extraocular muscle, lense, choroid plexus and viterous (white arrow) s/o pan ophthalmitis, ethmoid sinus show increased mucosal enhancement(solid arrow)

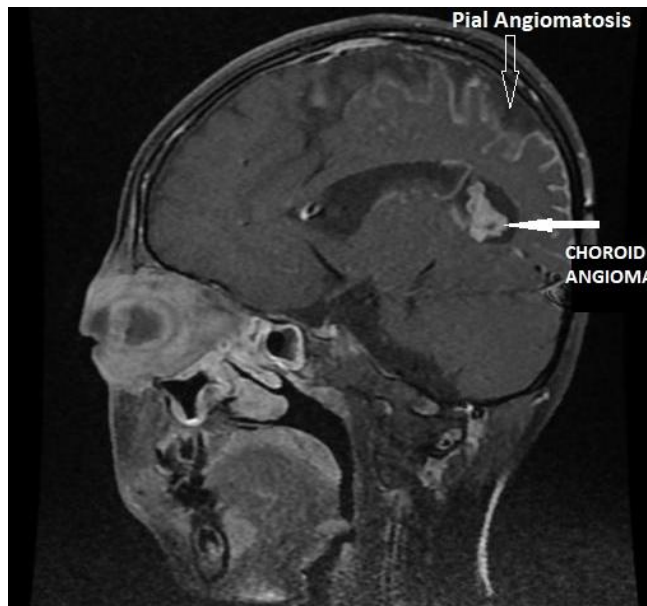


Image 5. Saggital contrast image depicts gyriform enhancemnt in the right parieto occipital region s/o pial angiomatosis (white arrow), choroid plexus appears enlarged and shows intense enhancement s/o choroidal angioma(solid white arrow)

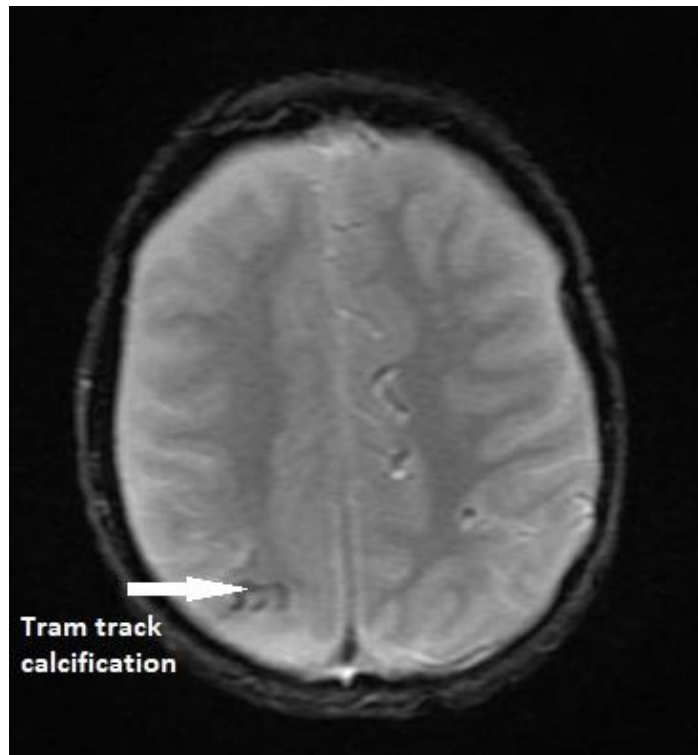


Image 6. Axial GRE image depicting linear areas of decreased altered signal intensity s/o areas of calcification

3. DISCUSSION

Phacomatoses also known as the neurocutaneous disorders are a wide and heterogenous group of disorders mainly effecting the structures which are derived from the neuro ectoderm such as –skin, eyes and the nervous system [6]. Sturge weber syndrome is also termed as encephalotrigeminal angiomasia or meningofacial angiomasia, is sporadic in nature in about 90% of the cases with presence of male dominance. The primary defect occurs in the 6th-8th week of gestation when there is defective formation of primordial vascular mesenchyme lying next to the neuro ectoderm, responsible for formation of head and face [7].

The most typical presentation is the port wine stains which occurs due to the capillary vascular malformation of the face, occurring in 90%of the patients, but few cases have been reported where there was absence of facial nevus and only intra cranial features like cerebral atrophy and pial angiomasia prevailed, these cases are labeled as atypical type of Sturge weber syndrome [1]. The second most common presentation occurring in 75-90% of the patients

is general or partial seizures, followed by developmental delay, about 1/3rd of the patients also suffer from hemiparesis as seen our patient.

The basic underlying pathophysiology for the neurological symptoms is the presence of pial angiomasia, due to the absence of adequate functioning of superficial cortical venous system. There is rerouting of blood via the medullary veins resulting in venous stasis and hypertension, this leads to abnormal venous drainage, progressive ischemia of underlying brain causing gradual cell death, loss of cerebral parenchyma. In chronic cases there is associated calcification of the cortex [4,7,8]. A number of related studies were reviewed [9-12].

The primary investigation done is a skull x ray which shows the typical tram track calcifications of the sulci and gyri, however these findings can be picked up on computer tomography. conventional radiography doesn't play any further role in diagnosing parenchymal and leptomenigeal involvement. MRI with post contrast gadolinium imaging stands as the investigation of choice for diagnosing neurocutaneous syndromes, the diagnostic

neuroimaging findings in a case of Sturge weber syndrome are as follows –

- i) Gyriformal pattern of leptomeningeal enhancement (due to venous congestion of internal cerebral veins) which signifies pial angiomatosis. This typical enhancement pattern is most commonly seen in parietal and occipital lobes of the supratentorial compartment [4].
- ii) Cerebral parenchymal atrophy with tram track calcifications noted on the side of pial angiomatosis. Few cases of progressive contralateral brain atrophy as seen in our case have been reported. Yoshikawa et al has described this phenomenon as crossed cerebral diaschisis (due to the functional disconnection of the cerebral cortex results in hypo metabolism of the contralateral cerebellar hemisphere) [8].
- iii) Other findings in neuroimaging include the presence of prominent and enlarged subependymal, deep medullary veins.
- iv) Enlarged Choroid plexus with increased enhancement is generally noted ipsilateral to the pial angiomatosis.
- v) Superior sagittal sinus thrombosis has been reported as an additional finding in few cases, primary seen in cases of bilateral Sturge weber syndrome due to stagnation of blood flow eventually forming a thrombus [1].
- vi) Abnormalities in the ipsilateral eye have been reported in Sturge weber syndrome, choroidal angiomas are seen as thickening of posterior aspect of globe with crescentic enhancement on post contrast images. As seen in our case there was presence of choroidal angioma with additional superadded findings of panophthalmitis which was possibly secondary to a foreign body injury to the eye.
- vii) Other ocular imaging findings includes choroidal and retinal detachments.

Early Radiological diagnosis of Sturge weber syndrome plays a crucial role in improving the patient prognosis. Making the diagnosis before the onset of clinical symptoms like seizure and mental retardation can help in better management of the patient, aggressive antiepileptic therapy, aspirin to prevent thromboembolisms should be started immediately [4]. The other mode of management in refractory cases include a focal brain resection and hemispherectomy.

4. CONCLUSION

Sturge weber syndrome is one of the rare neurocutaneous syndromes, where early diagnosis and prompt management can improve the patient's neurological prognosis, however SWS is not a static condition. The pathological changes taking place in the brain and dura leads to delayed diagnosis in patients having no or mild symptoms. Although CT has better efficacy in picking up intracranial calcifications, MRI with Gadolinium contrast is considered the gold standard in diagnosing pial angiomatosis, choroid plexus enlargement, prominent medullary veins and orbital choroid hemangiomas.

CONSENT

It is not applicable.

ETHICAL APPROVAL

It is not applicable.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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