A clinician's dilemma: Sturge-Weber syndrome 'without facial nevus'!!

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Abstract

Sturge–Weber syndrome (SWS) is a rare, sporadic neurocutaneous syndrome characterized by a classical triad of facial port wine nevus, ipsilateral leptomeningeal angiomatosis (LAM) and glaucoma. The incidence of SWS is 1/50,000 live births, although it is more often underreported. The incidence of SWS without facial nevus is not known, although very few patients without facial nevus have been reported. In these patients, the diagnosis of SWS is made by the findings of computed tomography, magnetic resonance imaging, and histopathology. Here, we report three patients with SWS from our cohort of 28 patients with SWS without facial nevus and discuss their clinical profile and outcome.

Key Words

Leptomeningeal angiomatosis, seizure, Sturge-Weber syndrome

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Introduction

Sturge–Weber syndrome (SWS) is a rare, sporadic neurocutaneous syndrome characterized by a classical triad of facial port wine nevus affecting the area innervated by the first sensory branch of the trigeminal nerve, ipsilateral leptomeningeal angiomatosis (LAM), and ocular involvement in the form of glaucoma.[1] The incidence of SWS is 1/50,000 live births, although it is more often underreported. It has been classified into three types, type I (facial and leptomeningeal angioma with possible glaucoma), type II (facial angioma without evident endocranial involvement), and type III (exclusive leptomengial angioma).[2] The incidence of SWS without facial nevus is not known, although few patients without facial nevus have been reported.[3-9] In these patients, the diagnosis of SWS has been made by the findings of computed tomography (CT), magnetic resonance imaging (MRI), and histopathology. Here, we report three patients with SWS without facial nevus from our cohort of 28 patients with SWS.

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Case series

Case 1

An 8-year-old boy presented with recurrent seizures since 5 years of age. He had normal birth and developmental history but poor scholastic performance. He had complex partial seizures (CPS) of extra temporal of left hemispheric origin with frequency of 3–4/month. He did not have any neurocutaneous markers, had impaired visual acuity (6/9 both eyes) with tubular field defect without evidence of glaucoma, left hemiatrophy of body, and left hemiparesis. His MRI showed right hemispheric atrophy with cortical T1/T2 hypointensity and intense blooming on susceptibility weighted images (SWI). Gadolinium enhanced T1 weighted images showed right temporo-occipital sulcal enhancement consistent with pial angiomatosis [Figures 1a-c]. A diagnosis of type III SWS was considered. On electroencephalography (EEG), right hemisphere showed voltage suppression with right posterior head region (PHR) spike wave discharges. He underwent right temporo-occipital lesionectomy in view of the refractory seizures and is seizure-free at 4 year follow-up. Histopathology showed vascular malformation with capillary telangectasia with extensive calcification supporting the diagnosis of SWS.

Case 2

A 7-year-old girl with normal birth and developmental history presented with recurrent CPS of occipital lobe semiology since 3 years of age. She did not have any neurocutaneous markers or any visual field defect or glaucoma. Her CT scan showed right

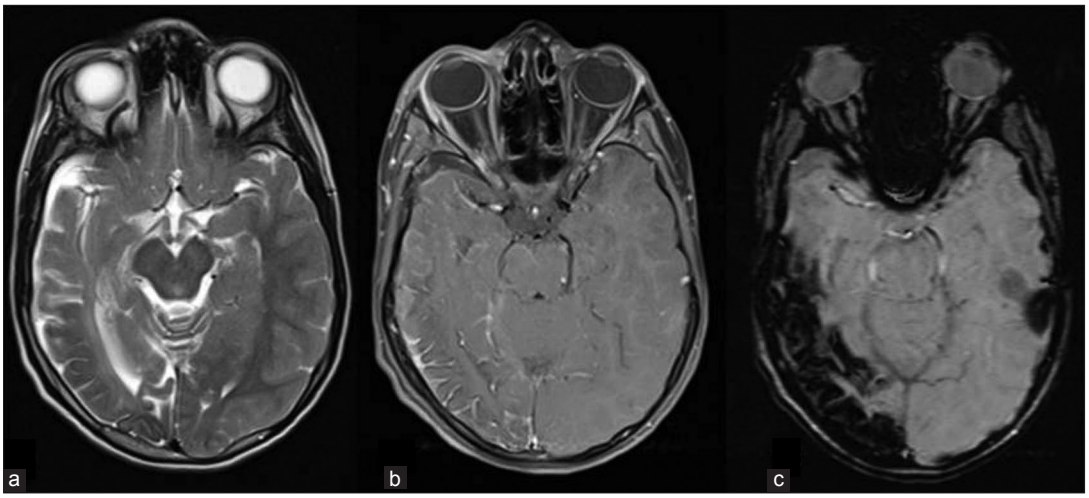

Figure 1: (a). T2 Weighted magnetic resonance image (MRI) showing right hemispheric atrophy with temporo-occipital cortical hypointensity, prominent sulcal spaces. (b). Gadolinium enhanced T1 weighted image showing right temporo-occipital sulcal enhancement consistent with pial angiomatosis. (c). Susceptibility weighted image (SWI) showing intense blooming of temporo-occipital cortex

temporo-parietooccipital calcification with right hemispheric atrophy. EEG showed right PHR spike wave discharges. Presently her seizures are well controlled on single antiepileptic drug at 10 month follow-up.

Case 3

A 23-year-old woman with normal birth and developmental history presented with recurrent right hemicranial throbbing headache with photophobia and phonophobia lasting 3-4 hours, relieved by vomiting and sleep, aggravated by travel and sleep deprivation since 21 years of age. She used to have a visual aura of flashes of light preceding the headache. The frequency of headache was 1–2/month. She had one episode of suspected transient loss of vision during one of the episode, so she underwent MRI head, which showed right occipital gyriform calcification typical of SWS. EEG was normal. She had left homonymous hemianopia without any neurocutaneous markers. She was initiated on flunarazine on which she had good control of her migraine at 6 year follow-up.

Discussion

The SWS is a neurocutaneous syndrome characterized by cutaneous facial angioma with angiomas involving the leptomeninges, glaucoma, seizures, stroke like episodes, hemiparesis, and mental retardation.[10] The classical SWS consist of triad of facial port wine stain in first trigeminal nerve distribution with LAM and glaucoma. In the absence of facial nevus, SWS can be diagnosed on the basis of clinical and radiological features. Very few patients with SWS without facial nevus have been reported world-over.[3-9] Seizures are the most common presentation of SWS, seen in 60–90% patients.[1] Out of our three patients without facial nevus, two presented with CPS and one with migraine. Seizure control is variable with some patients having refractory epilepsy, thought to be more common in patients who have bilateral nevus.[11] Age of onset of seizures is variable from early infancy to as late as third decade of life, onset during first year of life is thought to be associated with poor outcome.[12,13] One of our patients had refractory epilepsy requiring surgical treatment while the other was well controlled on medical management. Glaucoma is absent when nevus is not present in V1 distribution; therefore patients without nevus also may not have glaucoma. Mental retardation is seen in 30–50% patients of SWS and it is not related to presence or absence of facial nevus or extent of nevus as one of our patients had mental retardation.[1] Migraine is more common in SWS and is present in 30–50% patients, as one of our patients had migraine. The exact mechanism of migraine is not known, but is thought to be vascular in origin due to vasomotor changes in and around angioma.

Angioma in SWS is a low flow angioma with recurrent stasis and thrombosis, resulting in ischemia with brain injury resulting in gliosis. CT scan is usually the modality of choice in view of better delineation of calcification, which is seen as tram track calcification, however, it is usually absent in the early stages of the disease. MRI helps in the location and extent of LAM, underlying parenchymal abnormalities seen as hyperintensities (due to underlying gliosis). Susceptibility weighted sequence is useful in delineation of the mineral deposition and associated abnormal choroid plexus and venous drainage, which may be the only abnormality in some patients. The transmedullary collaterals develop in SWS secondary to insufficient superficial venous drainage.[14,15]

EEG shows suppression of voltage activity near the angioma and epileptiform discharges may rise from ipsilateral or contralateral hemisphere. The PHR most commonly shows epileptiform discharges followed by occipital lobe and then temporal, parietal, and frontal lobes with decreasing frequency, respectively. Some patients have frontal spikes, which can be explained by suppression of the background activity in the PHR.[16]

The diagnosis of SWS in patients without facial nevus who presents with neurological manifestations typical of SWS is made by the demonstration of leptomeningeal angioma

radiologically.[3,4] Cerebral calcifications are also seen in encephalitis, purulent meningitis, celiac disease, leukemia, and ossifying meningoencephalopathy, which should be excluded before diagnosis of SWS.[11] Histopathologic examination of SWS shows vascular malformation with capillary telangectasia with extensive calcification supporting the diagnosis of SWS.

The exact prevalence of SWS without facial nevus is not known; here we have reported three cases (10%) without facial nevus with neurological manifestations and radiological features classical of SWS with histopathologically proven diagnosis in one patient.

What this paper adds

- Sturge–Weber syndrome without facial nevus is one of the rare manifestations of the syndrome
- A very high index of clinical suspicion is needed to diagnose this entity which is often over looked

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