

Sturge-Weber Syndrome Without Facial Nevus

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Abstract

A patient with *Sturge-Weber* syndrome without the characteristic facial nevus presented with focal seizures which were difficult to control and borderline mental level. CT disclosed calcification in the right occipital zone. A marked decrease of the regional cerebral blood flow that extended beyond the abnormalities depicted on CT was seen by SPECT. Venous magnetic resonance (MR) angiography revealed reduction of the superficial cortical veins and prominent deep collateral venous system in the same side of the cerebral lesion. Cranial MR imaging with Gd-DTPA demonstrated the pial angioma.

Key words

CT – Epilepsy – MR – MR angiography – MR with Gd-DTPA – SPECT – *Sturge-Weber* syndrome

Introduction

Sturge-Weber syndrome (SWS) is a neuroectodermal disease characterized by a facial port-wine nevus that affects the area innervated by the first sensory branch of the trigeminal nerve, leptomeningeal angiomatosis ipsilateral to facial nevus, seizures of a mostly focal type, hemiparesis or hemiplegia which affects the contralateral side of leptomeningeal angiomatosis, and mental retardation. Diagnosis during life is usually made based on the presence of facial nevus flammeus and radiological evidence of intracranial calcification (1). A few cases of SWS without port-wine stain have been reported. Some were diagnosed on the basis of intracranial calcification (2) and some others were confirmed histologically and showed the leptomeningeal angiomatosis (6). Nowadays, however, magnetic resonance imaging (MRI) enhanced with gadolinium (Gd) DTPA discloses the cerebral and leptomeningeal lesions before the first evidence of SWS without needing histological confirmation for diagnosing this disease. A case with MRI with Gd-DTPA-enhanced MRI, MRI angiography and SPECT studies is described in this report.

Case report

The patient is a 20-year-old woman. She was the product of a fullterm uncomplicated pregnancy and delivery with a birth weight of 3700 grams. No abnormalities were noted during the first months of life. At 8 months of age the patient had a complex partial seizure of a duration of 5 minutes associated with fever. Within the following month, she presented four seizures of similar type unassociated with fever in spite of the antiepileptic medication (Phenobarbital, 4 mg/kg/day). Systemic neurologic examination was normal.

Electroencephalography (EEG), skull X-ray and pneumoencephalogram showed normal results. Phenytoin (3 mg/kg/day) was added to phenobarbital. Psychomotor development was normal and the patient was able to walk without support at 11 months of age and she started to speak at 13–14 months. She was free of seizures during about two years. Complex seizures reappeared at the age of 3 years. At this age, EEG showed focal spike activity in the right occipitoparietal region. Medication was changed to carbamazepine (20 mg/kg/day) but seizures were not controlled satisfactorily. At 6 years of age, X-ray of skull revealed calcific densities of SWS type in the right occipital area. Computed tomography (CT) scan presented a gyral pattern of cortical and subcortical calcification in the right occipital region. Phenytoin, carbamazepine, and valproate in various combinations controlled the seizures during some times although not completely. She, however, was able to live a normal life. She finished the primary studies at 15 years of age and started to work at 16 years.

At the age of 15 years, CT showed corticosubcortical calcifications in the right occipital zone with cortical atrophy in this area and enlargement of right choroid plexus. At 20 years of age, magnetic resonance (MR) revealed cortical and subcortical hypointensity of calcified leptomeningeal angioma in the right occipital zone. Enhanced MR with intravenous administration of gadolinium (Gd-DTPA) showed pial angiomatosis with adjacent cerebral angiomatosis in the right occipital area (Fig. 1) and enlargement and angiomatous appearance of the right choroid plexus. Venous MR angiography (MRA) revealed decrease of superficial cortical veins and prominent deep collateral venous system that drains into the cavernous sinus (Fig. 2). No changes were seen in retina. Arterial MRA did not present any anomaly. Single photon emission computed tomography (SPECT) with technetium Tc 99m (99mTc) showed a markedly hypoperfused area which extended on all the posterior zone of the right cerebral hemisphere beyond the area marked by the CT and the MR studies. Quantifying the perfusion on the symmetrical areas of both

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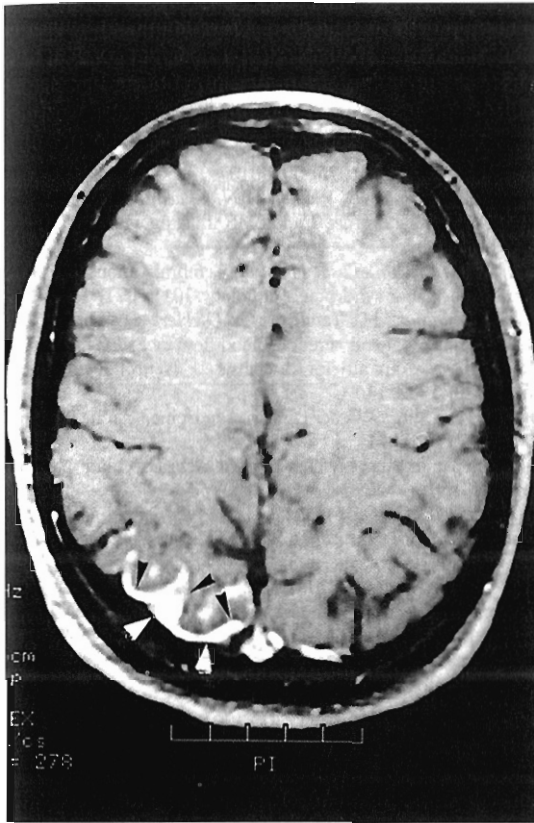


Fig. 1 Enhanced MR with Gd-DTPA discloses hypersignaled right occipital cortex-leptomeninges (arrowheads).

hemispheres revealed a valuable difference between them with decrease of the right one (Fig. 3).

Discussion

To accept as SWS cases without facial port-wine stains in spite of having other clinical and neuroradiological signs such as focal seizures, intracranial calcifications, hemiparesis and mental retardation usually has been a controversial subject (9). A high proportion of cases without facial port-wine nevus has been published in some series of SWS. *Peterman et al* (9) reported five of 35 patients, *Tönnis and Friedmann* (12) four of 23. *Lund* (7), however, found seven patients without the facial nevus with two proven at surgery in a review of 144 cases of SWS. This is more in agreement with our findings, one case without nevus and 40 with facial port-wine nevus (8). Ocular abnormalities are only present in patients of SWS with nevus flammeus affecting the upper eyelid (8). Because of this, there are no descriptions of SWS without port-wine nevus with ocular complications.

SPECT studies show from early ages decreased regional cerebral blood flow (rCBF) in the area of the lesion (5, 11).

Demonstration of pial angioma by imaging is only possible by MRI with Gd-DTPA (3, 8). It should be considered the most important criterion for the radiographic diagnosis of SWS (3) and it permits us to make the diagnosis of SWS without facial nevus flammeus during the first months of

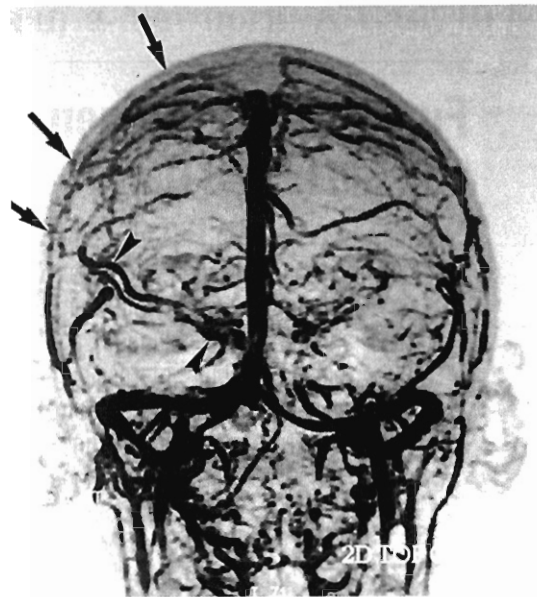


Fig. 2 Venous MRA in coronal image demonstrates decreased cortical veins in the right hemisphere (arrows) and the presence of a deep vein (arrowhead) with drainage into the cavernous sinus.

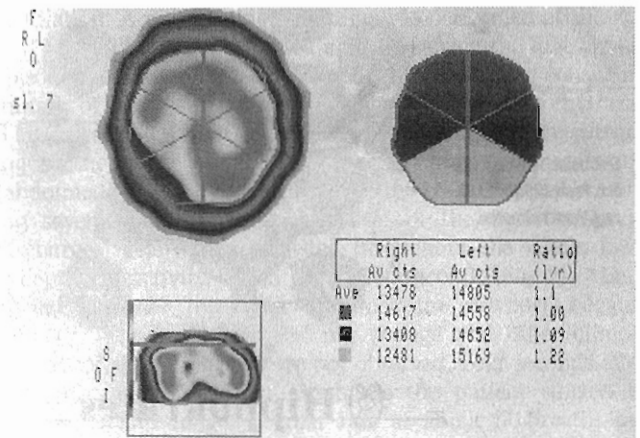


Fig. 3 Axial view of the technetium Tc 99m SPECT in quantified study showing a wide zone of hypoperfusion in the right occipital area. The table presents an important difference of perfusion between both parieto-occipital areas, severely decreased in the right side.

life. The lack of superficial cortical veins associated to thickening and tortuosity of the deep hemispheric veins ipsilateral to the facial nevus flammeus is also related to SWS (4, 10). To include or not cases without facial port-wine nevus as SWS in spite of showing intracranial lesions similar to those patients with facial angioma involving the forehead has been controversial. Nowadays, however, these cases are generally accepted as such. Perhaps in a next future molecular genetics may confirm or discard the diagnosis of SWS, maybe through the presence of some type of mosaicism.

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