Sturge-Weber syndrome, without a facial port-wine stain, with epilepsy onset in the fifth decade

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We describe a woman with SWS, without a facial nevus or eye abnormalities, and epilepsy onset at midlife.

Case report

A 46-year-old woman presented with a generalized seizure with the loss of consciousness. One prior episode had occurred three years earlier. There was no specific aura, and the episodes continued for about one minute. The patient had suffered from recurrent headaches that were sometimes accompanied by nausea since her twenties. Her family history was unremarkable.

The physical examination was normal and no facial port-wine stain was present. Neurological examination, including cognitive testing, revealed no abnormalities. A meticulous ophthalmological examination failed to reveal abnormalities such as raised intraocular pressure, heterochromia of the iris, choroidal hemangioma or conjunctival telangiectasia.

A routine electroencephalography was normal. An X-ray of the skull showed faint calcifications at the parieto-occipital area. A computed tomography (CT) scan of the brain revealed gyriform calcification with leptomeningeal angiomas in the left parieto-occipital area and atrophy of the left hemisphere (figure 1A). The brain magnetic resonance imaging (MRI) showed accentuation of leptomeningeal contrast enhancement involving the left parieto-occipital region (figure 1B).

The patient was treated with 200 mg of lamotrigine, and experienced no recurrence of the seizures during the following 12 months.

Discussion

SWS has been subdivided into three types: (1) type I - with facial, choroid and leptomeningeal angioma and possible glaucoma (classic form); (2) type II - with facial angioma without evident endocranial involvement; and (3) type III - with exclusive leptomeningeal angioma (Roach 1992). In our case, facial angioma and ocular manifestations were not present; according to this classification, she was considered to have SWS type III. Although the port-wine stain/nevus
flammeus is the pathognomic skin lesion present in 96% of cases (Kumar et al. 2004), leptomeningeal angiomatosis without a facial port-wine stain have been occasionally reported (Kumar et al. 2004, Comi et al. 2003, Aydin et al. 2000, Arzimanoglou et al. 2000, Maiuri et al. 1989). To our knowledge, this is the first report of SWS without facial nevus that presented with seizures in a middle-aged woman. It has been previously noted that the “atypical” cases of SWS, without a facial port-wine stain, may present later in childhood or young adulthood (Maiuri 1989). The reason for this difference from typical SWS is unclear but suggests that a less extensive insult to vascular development may result in milder impairment of brain development and function (Comi et al. 2003).

In the absence of facial angioma, the diagnosis is based on the radiological confirmation of the leptomeningeal angiomatosis and demonstration of the gyriform calcification on the CT scan. Enlargement and/or calcification of the choroid plexus, thickening of the calvarium and focal brain atrophy can also be observed (Gururaj et al. 2000). MRI may be more useful in detecting more subtle atrophic changes and enhanced MRI can effectively identify the abnormal brain and leptomeningeal vessels (Sen et al. 2002), even in the absence of calcifications. In our case, although no port-wine stain was found, the diagnosis of SWS was suggested by the calcifications noted on the skull X-ray and brain CT, and confirmed with contrast-enhanced MR images of the brain.

Seizures are the most common neurological manifestation and have been reported to occur in 23% to 83% of patients with SWS (Baselga 2004). The age of presentation of the seizures, and their evolution in SWS, is variable. In a comprehensive review of 171 personal cases, the age range for the onset of seizures varied between birth and 23 years, with a median of 6 months (Sujansky and Conradi 1995). SWS manifested as a seizure in a 45-year-old man, however, he had extensive, congenital port-wine stains on the face with chronic open-angle glaucoma (Hussain et al. 2004).

Epilepsy control in SWS is known to be difficult and usually requires the administration of more than one drug or even surgery (Arzimanoglou et al. 2000, Baselga 2004, Bourgeois et al. 2007). However, our patient became seizure-free with the usual dose of lamotrigine.

In summary, we report the first case of SWS, without a facial port-wine stain or ocular involvement, that presented with seizures in the fifth decade of life.

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