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turge-Weber syndrome or encephalofacial angiomatosis syndrome is a congenital and progressive disorder characterized by a port-wine stain in the area innervated by the first sensory branch of the trigeminal nerve, generalized or contralateral focal seizures as well as hemiplegia, homonymous hemianopsia, ipsilateral intracerebral calcifications, and mental retardation. Even though familial cases have been reported, it has not been proved that it is hereditary. Although classically, Sturge-Weber syndrome consist of unilateral facial nevus and ipsilateral intracerebral calcifications radiologically, atypical variants have been reported.2-4

DISSCUSSION

Classically, the diagnosis of Sturge-Weber syndrome is based on clinical and radiological findings.1,2 Unilateral facial nevus, and/or contralateral neurological deficits and seizures are among the clinical findings. In approximately 85% of cases, the blue-red colored nevus on the face is unilateral and always involves the upper part of the face and the eyelids. In the literature, Sturge-Weber syndrome cases without facial nevus have been reported,3 and cases with bilateral cutaneous involvement have been described.5 Bilateral facial angioma is seen in the literature, and its incidence has been determined as 15% of Sturge-Weber.5 Lindsey et al6 reported a case with bilateral facial and choroidal angioma. Bilateral choroidal angioma was reported as 15% among cases of Sturge-Weber syndrome in the study of Bebin et al7 and 25% in the study by Vilela.8 We detected both bilateral facial and choroidal angioma in our patient.

Choroidal angioma, buphthalmus, and glaucoma may be seen in the eye on the same side with facial angioma. Furthermore, megalocornea is very rarely detected in cases with Sturge-Weber syndrome.9 Interestingly, we detected megalocornea in the left eye of our case without other ophthalmic anomalies.

In cases with Sturge-Weber syndrome, neurological symptoms develop in months or years following birth. Seizures, which are generally the most frequently seen neurological finding, start before the age of 1 year. Generalized tonic-clonic seizures are the seizure type often seen in children. Infantile spasms, myoclonic and atomic seizures are among the less frequently seen seizures.1,10 In our case, focal seizures starting at the age of 2 months and probably secondary generalized seizures were observed later. Hemiparesis contralateral to the brain lesion may develop. Apart from this, mental retardation or learning difficulty may be seen. In our patient, even though there was no motor deficit, she was predominantly mentally and motor retarded in comparison with peers.

Sturge-Weber syndrome is diagnosed relatively easily by clinical and radiological findings. Intracerebral calcifications may be demonstrated by neuroimaging. In the cerebral CT, cortical calcifications are seen as a linear parallel “railroad-track” like configuration predominantly in the occipital and parieto-occipital regions. In our case, we saw calcifications predominantly in the bilateral frontal regions and extending to the bilateral temporo-parieto-occipital regions.

Based on seizures, neurological deficits, and radiological findings, the diagnosis of Sturge-Weber syndrome has been made in patients who have no facial nevus.3,11 In the cerebral CT, there are intracranial calcifications, enlargement in the choroidal plexus, and focal atrophy in the brain. The diagnosis of Sturge-Weber syndrome in our patient with a bilateral facial nevus who had typical clinical findings was made by radiological demonstration. In general, the most characteristic feature of Sturge-Weber syndrome is the presence of both ipsilateral facial nevus and intracranial calcifica-
what’s your diagnosis?

...tions. However, different variants of this syndrome have recently been reported in the literature. We thought that our case was interesting in having both bilateral facial nevus and intracranial calcifications and megalocornea without other ophthalmological abnormalities. Furthermore, we can suggest that the number of these variants will increase in the future and this may help in the delineation of underlying probable genetic defects and in creating new treatment modalities in Sturge-Weber syndrome.

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