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Pictorial Essay

Intracranial Manifestations of Tuberous Sclerosis: A Pictorial Essay

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Abstract:

Tuberous sclerosis is an autosomal dominant genetic disease that involves multiple organs. Hamartomas are the predominant lesions. Classically, tuberous sclerosis has been characterized by a classical clinical triad of facial angiofibromas (90%), mental retardation (50-80%), seizure (80-90%) and all three in 30% of the patients. Two major features or one major feature plus two minor features are necessary for the definite diagnosis of this disease. We had some patients admitted with different presentations of tuberous sclerosis and a past history of convulsion from childhood, skin lesions and also mental retardation with a new onset headache and a changed pattern of convulsion. In physical examination, facial angiofibromas and subungual fibromas were apparently detected. Brain CT scan study with contrast showed multiple calcified nodules associated with tubers, ventriculomegaly and also enhancing enlarged nodules at the foramen of Monro, which were suggestive of subependymal giant cell astrocytoma (SGCA). MRI showed the same brain findings (tubers, white matter lesions and subependymal nodules associated with SGCA), which were detected better. After surgery, SGCA was proved. In abdominal and pelvic CT scan and ultrasonography, massive bilateral angiomyolipomatosis and focal hypodense hyperechoic liver lesions were detected.

Keywords:

Tuberous Sclerosis Complex ، Tubers ، Subependymal Nodules ، Subependymal Giant Cell Astrocytoma ، White Matter Lesions

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