Neuroimaging of Tuberous Sclerosis (tubers-subependymal hamartomas)

6 years, female

Clinical History:
A 6 year-old female patient with seizures.

Imaging Findings:
Multiple intra-axial supratentorial lesions localized in corticosubcortical junction, in an approximate number of 16, of maximum size 9 mm, being iso-intense in T1 and T2 sequences, hyperintense in sequences in Flair, without restriction in diffusion, without significant contrast uptake.
Multiple subependymal lesions distributed in the lateral ventricles, especially anterior horns, hyperintense in T1, hypointense in T2 and Flair, with moderate enhancement after administration of intravenous contrast.

Discussion:
Tuberous sclerosis, also called Tuberous Sclerosis Complex, included in neurocutaneous disorders with multiorganic hamartomas.
Tuberous sclerosis the majority being sporadic (85%) in an autosomal dominant fashion (15%). Caused by mutation in TSC1 or TSC2 gene[1, 2, 5]

Clinical Perspective: There is a classic clinical triad; Facial angiofibromas (90%), mental retardation (50-80%), seizures (80-90%).

Image Perspective: The manifestations of tuberous sclerosis in the brain are: Tubers, subependymal nodes, white matter abnormalitie, subependymal astrocytoma of giant cells (SGCAs), etc.
Tubers: They are benign hamartomatous lesions with epileptogenic potential at cortical level they occur in (95-100%) of the cases and up to 90% are located in the frontal lobes. In T1-WI, they are hypointense and hyperintense in T2-Flair-WI, demonstrating enhancement (5%).
Subependymal nodules: small hamartomas on the inner walls of the lateral ventricles, which can converge with each other. They are found most frequently in the caudothalamic sulcus in the foramen region of Monro. Being hyperintense in T1-WI and iso and hyperintense in T2-weighted images, presenting enhancement in 3-4% of the
cases. They are benign, but one (5-10%) can degenerate into SGCAs. Suspicion for transformation should be considered if lesions measure 5 mm or greater in diameter, are incompletely calcified, and demonstrate enhancement. The enhancement is variable and can not be used to differentiate (SGCAs), being more useful long-term growth for this purpose. [1, 2, 3, 4]

White matter abnormalities: They include superficial abnormalities associated with tubers, radial lines, lesions similar to cysts. In MR are seen as hyperintense in T2-WI and hypointense in T1-WI. The radial migration lines refers to the linear bands observed in MRI that range from the periventricular white matter to the subcortical region. These radial migration lines often end in a tuber.

Subependymal giant cell astrocytomas: benign tumors that are found almost exclusively in tuberous sclerosis (5-15%). His presence is virtually pathognomonic. The classic location is close to the foramen of Monro and they develop from a subependymal node. In CT images they are iso or slightly hypodense in relation to the gray matter, often with associated cacominations. Show heterogenous enhancement on MR imaging, demonstrating T1 isointense and hypointense signal and T2/FLAIR isointense and hyperintense signal.[1, 2, 3, 4]

Treatment: the control of the convultions and in case of being refractory the surgical excision of the tubercle can be realized. In the case of the (SGCAs) the treatment is surgery, and immunosuppressive therapy (Sirolimus).[1, 2, 3, 4]

"Written informed patient consent for publication has been obtained"

**Differential Diagnosis List:** Tuberous sclerosis (Bourneville disease), TORCH, Neurocysticercosis, Tuberculoid granuloma, Taylor-Type Cortical Dysplasia, X-Linked Subependymal Heterotopia

**Final Diagnosis:** Tuberous sclerosis (Bourneville disease)

**References:**


Figure 1

Description: Axial T2 Flair WI MR. Several in corticosubcortical junction intra-axial hyperintense lesions (tubers) are observed. Origin: Department of Radiology, Hospital Universitario Rio Hortega, Valladolid, Spain
Description: Axial T2 Flair WI MR. Several in corticosubcortical junction intra-axial hyperintense lesions (tubers) are observed. Origin: Department of Radiology, Hospital Universitario Rio Hortega, Valladolid, Spain
Description: Coronal T2 Flair WI MR. Several in corticosubcortical junction intra-axial hyperintense lesions (tubers) are observed. Origin: Department of Radiology, Hospital Universitario Rio Hortega, Valladolid, Spain
Figure 2

Description: Axial T2 WI MR. A subependymal nodule in the frontal horn of the left lateral ventricle, adjacent to the foramen of Monro, iso-intense in relation to the surrounding parenchyma. Origin: Department of Radiology, Hospital Universitario Rio Hortega of Valladolid, Spain.
Description: Axial T1 C+ WI MR. A subependymal nodule in the frontal horn of the left lateral ventricle, adjacent to the foramen of Monro, with important enhancement. Origin: Department of Radiology, Hospital Universitario Rio Hortega of Valladolid, Spain.
Description: Coronal T1 C+ WI MR. Several subependymal nodular lesions are observed along the walls of both lateral ventricles, with important enhancement. Origin: Department of Radiology, Hospital Universitario Rio Hortega of Valladolid, Spain.