Tuberous sclerosis

Tuberous sclerosis (TSC) is an autosomal dominant, multi-organ disorder characterized by the development of hamartomas and benign tumors in the brain, heart, kidneys, liver, lungs, and other sites.

Diagnostic criteria according to the International Tuberous Sclerosis Complex Consensus conference are listed here:

Major features

1. Hypomelanotic macules (≥3, at least 5 mm in diameter)
2. Angiofibromas or fibrous forehead plaque
3. Ungual or periungual fibroma (>2)
4. Shagreen patch (connective tissue nevus)
5. Multiple retinal hamartomas
6. Cortical dysplasias (≥3, including tubers and cerebral white matter migration lines)
7. Subependymal nodules (≥2)
8. Subependymal giant cell astrocytoma (SEGA)
9. Cardiac rhabdomyoma
10. Lymphangiomyomatosis
11. Renal angiomyolipoma

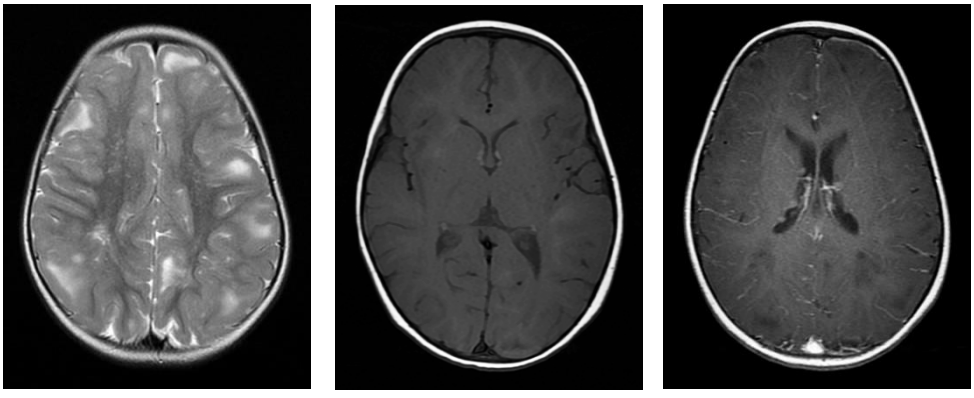
Minor features

1. Dental enamel pits (≥3)
2. Intraoral fibromas (≥2)
3. Nonrenal hamartoma (histologic confirmation suggested)
4. Retinal achromic patch
5. “Confetti” skin lesions
6. Multiple renal cysts

Definite diagnosis: at least two major features or one major and two minor criteria
Possible diagnosis: either one major or ≥2 minor features

The most common clinical manifestations of TSC include epilepsy and cognitive impairment. More recent studies have shown a predisposition of some patients with TSC to develop autism.

Our 2-year old patient with hypopigmented macules (#1 major criterion) presented with seizures. Axial T2-weighted image (leftmost picture) demonstrates hyperintense cortical tubers (#6 major criterion). Pre- and post-contrast axial T1-weighted images (middle and rightmost pictures, respectively) shows multiple hyperintense and enhancing subependymal nodules (#7 major criterion). Having three of the major criteria fulfill the diagnosis of Tuberous Sclerosis.



Tubers

Tubers (cortical hamartomas) are most commonly found in the cerebral hemispheres. Clinical features which depend on the location include abnormalities in cognition, cranial nerve deficits, focal motor or sensory abnormalities, cerebellar dysfunction, and gait abnormalities.

The appearance of cortical tubers on MRI changes with age. In neonates, they appear as hyperintense gyri on T1W and are hypointense to white matter on T2W. This signal abnormality may extend through the cortical mantle to the ventricular surface. The lesions gradually become isointense as myelination progresses. In older infants, the tubers are hypointense on T1W, with cortical and subcortical areas of T2 and FLAIR hyperintensity. They may rarely develop calcification, and a small proportion of degenerated, calcified cortical tubers (3-4%) will enhance following contrast administration.

Subependymal nodules

Subependymal nodules (SENs) are benign hamartomatous nodules found along the walls of the lateral ventricles, most commonly at the caudothalamic groove just posterior to the foramen of Monro. They are typically benign lesions. Subependymal nodules can enlarge and degenerate into Subependymal Giant Cell Astrocytomas (SEGAs) in 10-20% of cases. Development of SEGAs is a gradual process that usually occurs within the first two decades of life.

SENs appear echogenic on cranial US and can be difficult to differentiate from periventricular gray matter heterotopia. Imaging appearances on CT and MRI also change with age. In the first few years of life, the number of calcified lesions gradually increases. They are T1 hyperintense and T2 hypointense in young infants with unmyelinated white matter. As myelination progresses, SENs become more isointense to white matter and best visualized on T1W, where their hyperintensity contrasts with the low signal intensity of CSF. Larger nodules may show variable degrees of T2 hypointensity related to the presence of calcification. T2* MR images are ideal to demonstrate the presence of calcification.

Both SEGAs and subependymal nodules enhance following contrast administration. The most useful distinguishing imaging feature is growth on serial imaging studies.

Source: Excerpts were taken from Pediatric Radiology: Practical Imaging Evaluation of Infants and children. Edward Y. Lee, editor. Wolters Kluwer 2018, pp. 151-152

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