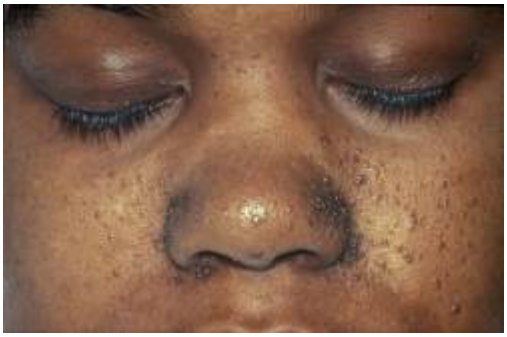
Tuberous sclerosis complex



Tuberous sclerosis complex, composed of the Latin tuber (swelling) and the Greek skleros (hard), refers to the pathological finding of thick, firm and pale gyri, called “tubers,” in the brains of patients postmortem. These tubers were first described by Désiré-Magloire Bourneville in 1880; the cortical manifestations may sometimes still be known by the eponym Bourneville's disease.

Tuberous sclerosis complex (TSC) is an autosomal dominant multisystem disease usually diagnosed in childhood.

Tuberous sclerosis complex (TSC), AKA Bourneville’s disease, is a neurocutaneous disorder characterized by hamartomas of many organs including the skin, brain, eyes and kidneys. In the brain, the hamartomas may manifest as cortical tubers, glial nodules located subependymally or in deep white matter, or giant cell astrocytomas. Associated findings include pachygyria or microgyria.

Tuberous sclerosis complex (TSC) was initially described approximately 150 years ago by von Recklinghausen in 1862 [1].

Subependymal giant cell astrocytomas (SEGA) are benign brain lesions occurring in up to 20% of patients with TSC.

Key concepts

● most cases are due to spontaneous mutation. Inherited cases are autosomal dominant. Incidence: 1 in 6K–10K live births.

● classic clinical triad: seizures, mental retardation, and sebaceous adenomas; the full clinical triad is seen in < 1/3 of cases.

● typical CNS finding: subependymal nodules (“tuber”)—a hamartoma.

● commonly associated neoplasm: subependymal giant cell astrocytoma (SEGA)

● 2 tumor suppressor genes: TSC1 (on chromosome 9q34) codes for hamartin and TSC2 (on chromosome 16p13) encodes tuberin

● CT shows intracerebral calcifications (usually subependymal).

Epidemiology

Studies estimate a frequency of 1/6000 to 1/10,000 live births and a population prevalence of around 1 in 20,000 [2) 3)].

Etiology

Tuberous sclerosis complex etiology.

Clinical features

Tuberous sclerosis complex clinical features

Diagnosis

Tuberous sclerosis complex diagnosis.

Pathology

Subependymal nodules (“tubers”) are benign hamartomas that are almost always calcified, and protrude into the ventricles.

▶ Subependymal giant cell astrocytoma (SEGA). Almost always located at the foramen of Monro. Occurs in 5–15% of patients with TSC.

Treatment

Tuberous sclerosis complex treatment.

Outcome

Tuberous sclerosis complex prognosis.

Case series

Tuberous sclerosis complex case series.

Case reports

Lv H, Wang X. Space-occupying Intraventricular Vascular Lesion in Tuberous Sclerosis Complex. Neurologist. 2022 Jan 26. doi: 10.1097/NRL.0000000000000415. Epub ahead of print. PMID: 35081607 [4].

A novel technique is presented for the application of MRgLITT in a 6-month-old infant for the treatment of epilepsy associated with tuberous sclerosis complex (TSC) .

To Hooten et al. from the Tuberous Sclerosis Complex Clinic, Duke University, Durham, North Carolina; and University of Florida, Gainesville, knowledge this is the youngest patient treated with laser ablation. They used a frameless navigation technique with a miniframe tripod system and intraoperative reference points. This technique expands the application of MRgLITT to younger patients, which may lead to safer surgical interventions and improved outcomes for these children [5].

References

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von Recklinghausen F. Die Lymphelfasse und ihre Beziehung zum Bindegewebe. [German]. Berlin: A. Hirschwald; 1862.
2)
O'Callaghan F, Shiell A, Osborne J, Martyn C. Prevalence of tuberous sclerosis estimated by capture-recapture analysis. Lancet. 1998;352:318–319.
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Sampson J, Scahill S, Stephenson J, Mann L, Connor J. Genetic aspects of tuberous sclerosis in the west of Scotland. J Med Genet. 1989;26:28–31.
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Hooten KG, Werner K, Mikati MA, Muh CR. MRI-guided laser interstitial thermal therapy in an infant with tuberous sclerosis: technical case report. J Neurosurg Pediatr. 2018 Sep 28:1-6. doi: 10.3171/2018.6.PEDS1828. [Epub ahead of print] PubMed PMID: 30265228.