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NLM Citation: National Center for Biotechnology Information (US). Genes and Disease [Internet]. Bethesda (MD): National Center for Biotechnology Information (US); 1998-. Tuberous sclerosis.
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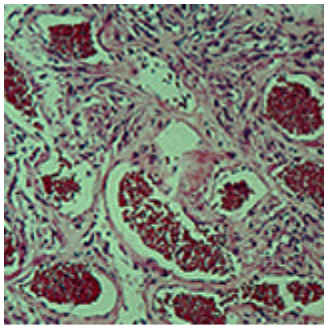
Tuberous sclerosis



Microscopic section of angiomyolipoma, a benign tumor of the kidney present in many patients with tuberous sclerosis. [Image credit: Moyra Smith, Johns Hopkins University, Baltimore, MD, USA.]

Tuberous sclerosis is an hereditary disorder characterized by benign, tumor-like nodules of the brain and/or retinas, skin lesions, seizures and/or mental retardation. Patients may experience a few or all of the symptoms with varying degrees of severity.

Two genes for tuberous sclerosis have been found: TSC1 on chromosome 9, and TSC2 on chromosome 16. It took four years to pin down a specific gene from the TSC1 region of chromosome 9: in 1997, a promising candidate was found. Called hamartin by the discoverers, it is similar to a yeast protein of unknown function, and appears to act as a tumor suppressor: without TSC1, growth of cells proceeds in an unregulated fashion, resulting in tumor formation. TSC2 codes for a protein called tuberin, which, through database searches, was found to have a region of homology to a protein found in pathways that regulate the cell (GAP3, a GTPase-activation protein).

TSC1 has a homolog in yeast, which provides a system in which to model the human disease.

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