Tuberous Sclerosis Complex: An Overview

Approximately **50,000** people in the United States have TSC[1]

TSC occurs in all races and ethnic groups, and in both genders[2]

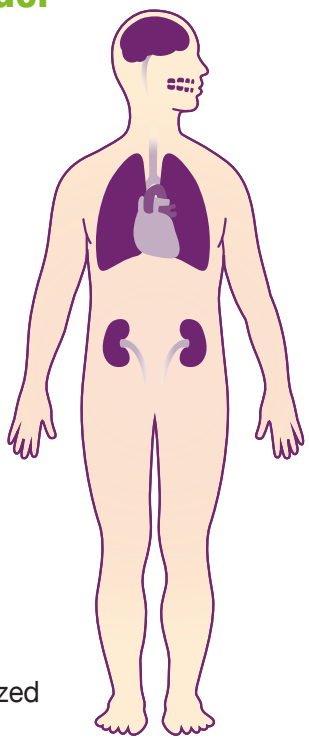
The disease affects an estimated **1 in 6,000** newborns[2]

Diseases with similar US prevalence rates include cystic fibrosis (approximately 30,000 people) and amyotrophic lateral sclerosis (ALS), or Lou Gehrig's disease (up to approximately 30,000 people)[3,4]

About **1/3** of all people with TSC genetically inherit the disease, while in the remaining individuals, the disease is acquired as a result of spontaneous genetic mutation[5]

Tuberous Sclerosis Complex (TSC) is a Multiorgan Genetic Disorder

- It is characterized by the formation of hamartomas, which are noncancerous tumor-like masses[2,5]
- These tumors can form in major organs including the brain, skin, eyes, and kidneys. Tumors in the heart often occur in children, while lung tumors can occur in adults[2,5,6]
- Symptoms of TSC can range from mild to severe and can change over time[2,6]
  - TSC may not be noticeable. Because symptoms vary and may not be immediately recognized by a health care provider, TSC is often undiagnosed for years

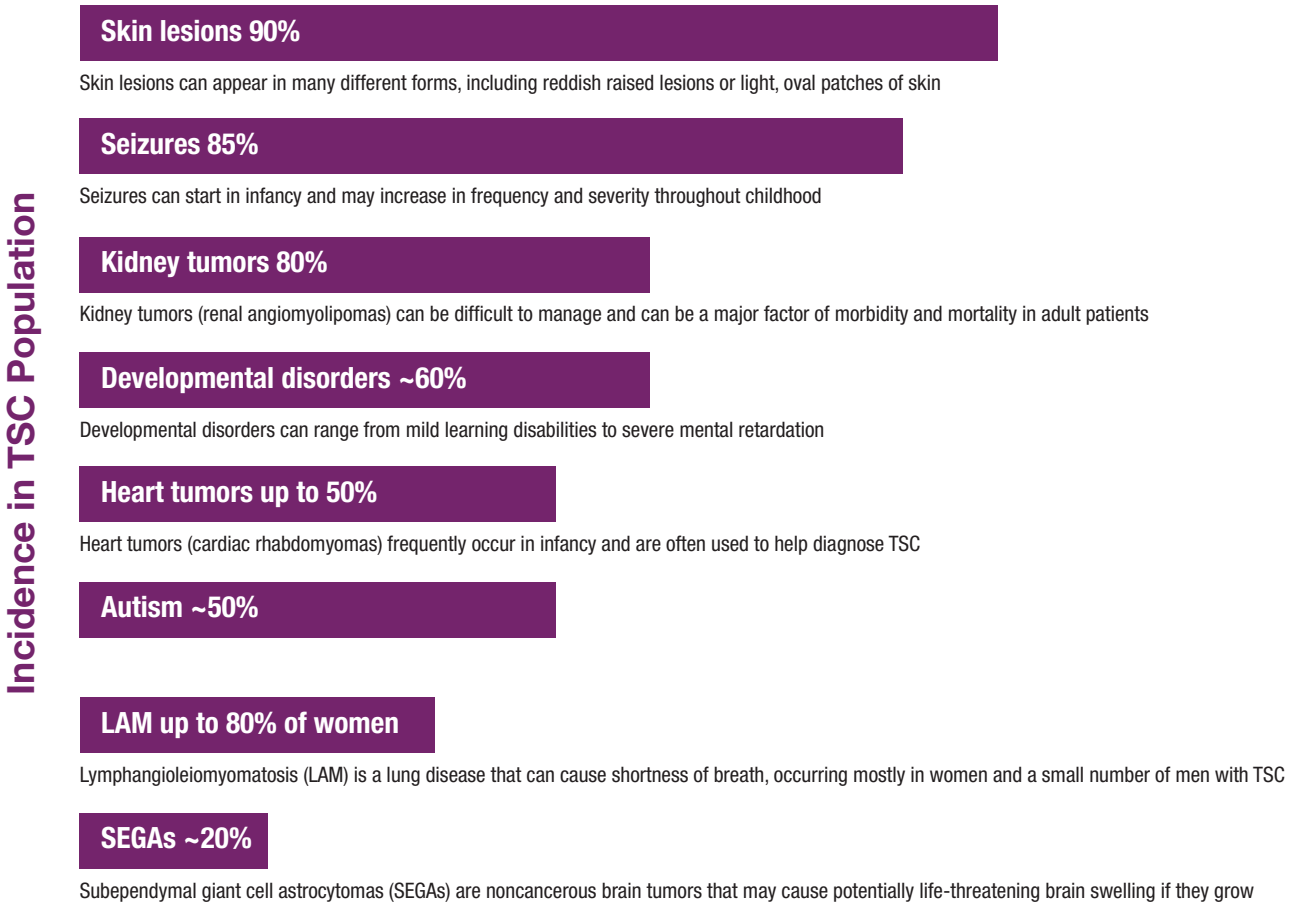


Depending on the body organs affected by TSC, different specialists may be involved, such as a:

Nephrologist or *urologist*	for kidney manifestations, such as renal angiomyolipoma
Neurologist	for brain manifestations, such as subependymal nodules (SENs) and subependymal giant-cell astrocytomas (SEGAs)
Dermatologist	for skin manifestations
Pulmonologist	for lung complications, such as lymphangioleiomyomatosis (LAM)
Psychiatrist or *psychologist*	for issues related to cognition, mood, and behavior disorders

TSC affects multiple organs

TSC SIGNS, SYMPTOMS AND RESULTING DISORDERS[6-8]



References: 1. Wu EW, Kirov II, Tal A, et al. Brain MR spectroscopic abnormalities in "MRI-negative" tuberous sclerosis complex patients. *Epilepsy & Behavior.* 2013;(27):319-325. **2.** National Institutes of Health: National Institute of Neurological Disorders and Stroke. Tuberous Sclerosis fact sheet. http://www.ninds.nih.gov/disorders/tuberous_sclerosis/detail_tuberous_sclerosis.htm. Accessed February 18, 2015. **3.** Genetic Disease Profile: Cystic Fibrosis. www.genomics.energy.gov. Accessed February 19, 2015. **4.** National Institutes of Health: National Institute of Neurological Disorders and Stroke. ALS (Amyotrophic Lateral Sclerosis) fact sheet. http://www.ninds.nih.gov/disorders/amyotrophiclateralsclerosis/detail_ALS.htm. Accessed February 18, 2015. **5.** Curatolo P, Verdecchia P, Bombardieri R. Tuberous sclerosis complex: A review of neurological aspects. *Eur J Paed Neurol.* 2002;6:15-23. **6.** Northrup H, Krueger DA, on behalf of the International Tuberous Sclerosis Complex Consensus Group. Tuberous sclerosis complex diagnostic criteria update: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. *Pediatr Neurol.* 2013;49:243-254. **7.** Wataya-Kaneda M, Tanaka M, Hamasaki T, Katayama I. Trends in the prevalence of tuberous sclerosis complex manifestations: An epidemiological study of 166 Japanese patients. 2013: PLoS ONE 8(5): e63910. **8.** Thiele EA. Managing and understanding epilepsy in tuberous sclerosis complex. *Epilepsia.* 2010;51 (suppl. 1):90–91.

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