

# Tuberous Sclerosis

## Complex: An Overview



Approximately  
**50,000**  
people in the United States  
have TSC<sup>1</sup>



TSC occurs in all races  
and ethnic groups, and in  
both genders<sup>2</sup>



The disease  
affects an estimated  
**1 in 6,000**  
newborns<sup>2</sup>



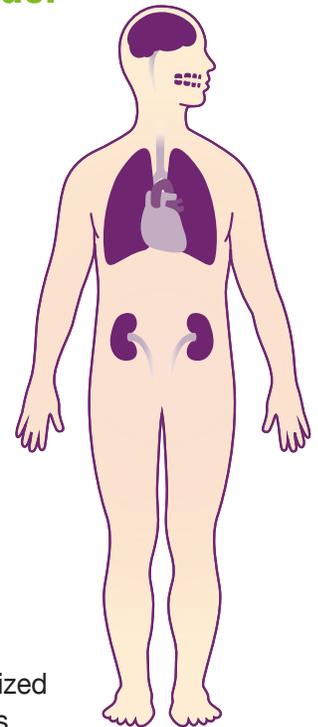
Diseases with similar  
US prevalence rates  
include cystic fibrosis  
(approximately 30,000  
people) and amyotrophic  
lateral sclerosis (ALS),  
or Lou Gehrig's disease  
(up to approximately  
30,000 people)<sup>3,4</sup>



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<sup>5</sup>

## Tuberous Sclerosis Complex (TSC) is a Multiorgan Genetic Disorder

- It is characterized by the formation of hamartomas, which are noncancerous tumor-like masses<sup>2,5</sup>
- 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<sup>2,5,6</sup>
- Symptoms of TSC can range from mild to severe and can change over time<sup>2,6</sup>
  - 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:



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

# TSC affects multiple organs

## TSC SIGNS, SYMPTOMS AND RESULTING DISORDERS<sup>6-8</sup>

Incidence in TSC Population

### Skin lesions 90%

Skin lesions can appear in many different forms, including reddish raised lesions or light, oval patches of skin

### Seizures 85%

Seizures can start in infancy and may increase in frequency and severity throughout childhood

### Kidney tumors 80%

Kidney tumors (renal angiomyolipomas) can be difficult to manage and can be a major factor of morbidity and mortality in adult patients

### Developmental disorders ~60%

Developmental disorders can range from mild learning disabilities to severe mental retardation

### Heart tumors up to 50%

Heart tumors (cardiac rhabdomyomas) frequently occur in infancy and are often used to help diagnose TSC

### Autism ~50%

### LAM up to 80% of women

Lymphangiomyomatosis (LAM) is a lung disease that can cause shortness of breath, occurring mostly in women and a small number of men with TSC

### SEGAs ~20%

Subependymal giant cell astrocytomas (SEGAs) are noncancerous brain tumors that may cause potentially life-threatening brain swelling if they grow

**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](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](http://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](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.