

Tuberous Sclerosis Complex: An In-Depth Exploration of Genes, Clinical Features, and Therapeutic Approaches

Tuberous Sclerosis Complex (TSC) Patient Journey

What is TSC?

Tuberous sclerosis complex (TSC), is a genetic disorder that may cause non-cancerous tumors to form in vital organs and can affect many parts of the body, including the brain, kidneys, heart, lungs and skin, with some patients exhibiting seizures, autism, cognitive delays or behavior issues. TSC is a highly variable condition, with no two people experiencing the same symptoms or disease progression.

As manifestations vary from person to person, the onset of TSC can take years to develop and it may go undetected, with many children not diagnosed until later in life. Because TSC is a lifelong condition, individuals need to be monitored regularly by a doctor experienced with the disorder to ensure tumor growth or new symptoms are identified early.

TSC affects up to **1 MILLION** people worldwide

Approximately **50,000** people in the US have TSC

TSC affects an estimated **1 in 6,000** newborns. TSC occurs in all races and ethnic groups, and in both genders.

Organ System	Condition	Prevalence
Heart Tumors	Cardiac Rhabdomyomas	~50% of newborns with TSC
	Retinal Nodules (hamartomas)	40-50% of people with TSC
Brain Lesions	Subependymal Nodules (SEN)	~90% appearance rate in people with TSC
	Cortical Tubers	>80% appearance rate in people with TSC
Skin Lesions	Hypomelanotic Macules (3 or more)	>90% of people with TSC
	Forearm Angiokeratomas	~75% of people with TSC
Neurological Manifestations	Seizures Associated with TSC	~85% of people with TSC are affected by epilepsy
	Behavioral Disorders	~60% of people with TSC fail to demonstrate seizure control available treatments
Lung Disease	Lymphangioma-leiomyomatosis (LAM)	<40% of women with TSC
	Periungual or Ungual Fibromas	<12% of men with TSC
Fingernail & Toenail Tumors	Periungual or Ungual Fibromas	~50% of people with TSC
	Brain Tumors	~20% of people with TSC
Brain Tumors	Subependymal Giant Cell Astrocytomas (SEGAs)	~60% of people with TSC
	Brain Tumors	~20% of people with TSC

Stages of Human Life

FETUS: Cardiac Rhabdomyomas, Retinal Nodules (hamartomas)

INFANT: Subependymal Nodules (SEN), Cortical Tubers

ADOLESCENT: Hypomelanotic Macules (3 or more), Forearm Angiokeratomas, Seizures Associated with TSC, Behavioral Disorders, Skin Lesions

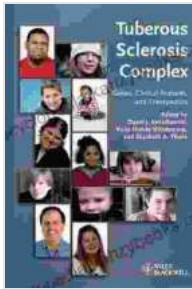
ADULT: Lymphangioma-leiomyomatosis (LAM), Periungual or Ungual Fibromas, Brain Tumors, Subependymal Giant Cell Astrocytomas (SEGAs)

Note: Manifestations are included above at the point during human development when the probability is known to first occur in a patient. Behavioral manifestations include the stages of life during which a manifestation is known to occur. Age-specific prevalence statistics are available for certain manifestations.

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Tuberous Sclerosis Complex (TSC) is a rare genetic disorder characterized by the formation of benign tumors in multiple organs,

including the brain, kidneys, heart, lungs, and skin. It is caused by mutations in either the TSC1 or TSC2 gene, which are responsible for encoding proteins involved in the regulation of cell growth and proliferation. TSC affects an estimated 1 in 6,000 individuals worldwide, with equal distribution between males and females.



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Genetic Basis of TSC

TSC is an autosomal dominant disorder, meaning that the affected gene is located on one of the non-sex chromosomes. Mutations in either the TSC1 or TSC2 gene can cause TSC. These mutations result in the production of defective proteins, which leads to the uncontrolled growth of cells and the formation of tumors.

Approximately two-thirds of TSC cases are caused by mutations in the TSC2 gene, located on chromosome 16. The remaining one-third of cases are caused by mutations in the TSC1 gene, located on chromosome 9. Mutations in these genes lead to the disruption of the TSC1-TSC2

complex, which plays a critical role in regulating cell growth and proliferation.

Clinical Features of TSC

TSC can manifest in a wide spectrum of clinical features, depending on the severity of the mutations and the organs affected. The most common clinical features include:

- **Neurological features:** Seizures, autism spectrum disorder, intellectual disability, behavioral problems, developmental delays
- **Renal features:** Renal cysts, angiomyolipomas (benign kidney tumors), end-stage renal disease
- **Cardiac features:** Cardiac rhabdomyomas (benign heart tumors), arrhythmias, heart failure
- **Pulmonary features:** Lymphangiomyomatosis (LAM), a rare lung disease that affects women
- **Skin features:** Facial angiofibromas (small red bumps on the face), hypomelanotic macules (white spots on the skin)
- **Ocular features:** Retinal hamartomas (benign tumors in the retina), glaucoma
- **Dental features:** Enamel pits, intraoral fibromas

Diagnosis of TSC

TSC is typically diagnosed based on a combination of clinical features, physical examination findings, and genetic testing. Magnetic resonance imaging (MRI) and computed tomography (CT) scans are often used to

visualize the tumors in different organs. Genetic testing can confirm the diagnosis of TSC by identifying mutations in the TSC1 or TSC2 genes.

Treatment of TSC

There is currently no cure for TSC, but treatment focuses on managing the symptoms and preventing complications. Treatment options may include:

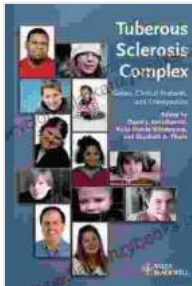
- **Medical management:** Medications to control seizures, prevent kidney and heart problems, and treat LAM
- **Surgical intervention:** Surgery to remove or reduce the size of tumors in the brain, kidney, heart, or lungs
- **Laser therapy:** To remove facial angiofibromas
- **Psychotherapy:** To address behavioral and developmental issues
- **Regular monitoring:** Regular check-ups with specialists to monitor the progression of the disease and detect any potential complications

Prognosis of TSC

The prognosis for individuals with TSC varies depending on the severity of the mutations and the organs affected. The most common cause of death in TSC is complications related to the brain tumors, such as seizures and epilepsy. However, with early diagnosis and proper management, most individuals with TSC can live full and productive lives.

Tuberous Sclerosis Complex (TSC) is a complex genetic disorder that can affect multiple organs. The clinical manifestations of TSC vary widely, but early diagnosis and proper management are crucial for improving outcomes. Ongoing research is focused on understanding the

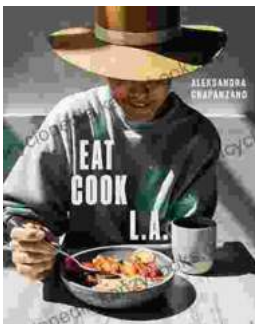
genetic basis of TSC, developing new treatments, and improving the quality of life for individuals affected by this condition.



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