

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 individuals worldwide
- Approximately 50,000 people in the US have TSC
- TSC occurs in all races and ethnic groups, and in both genders
- TSC affects an estimated 1 in 6,000 newborns



Neurological Manifestations

Seizures Associated with TSC
 ~85% of people with TSC are affected by epilepsy
 ~60% of people with TSC fail to demonstrate seizure control with available therapies

Behavioral Disorders
 Mental Disabilities
 ~50% of people with TSC exhibit autistic characteristics

Brain Tumors
 Subependymal Giant Cell Astrocytomas (SEGAs)
 ~20% of people with TSC



Kidney Tumors

Renal Angiomyolipomas
 ~60% of people with TSC



Fingernail & Toenail Tumors

Periungual or Ungual Fibromas
 ~20% of people with TSC



Lung Disease

Lymphangiomyomatosis (LAM)
 ♀ <40% of women with TSC
 ♂ <12% of men with TSC



Eye Tumors

Retinal Nodular Hamartomas
 40-50% of people with TSC



Skin Lesions

>95% of people with TSC
 Hypomelanotic Macules (3 or more)
 >90% of people with TSC
 Facial Angiofibromas
 ~75% of people with TSC
 Shagreen patches
 ≤30% of people with TSC



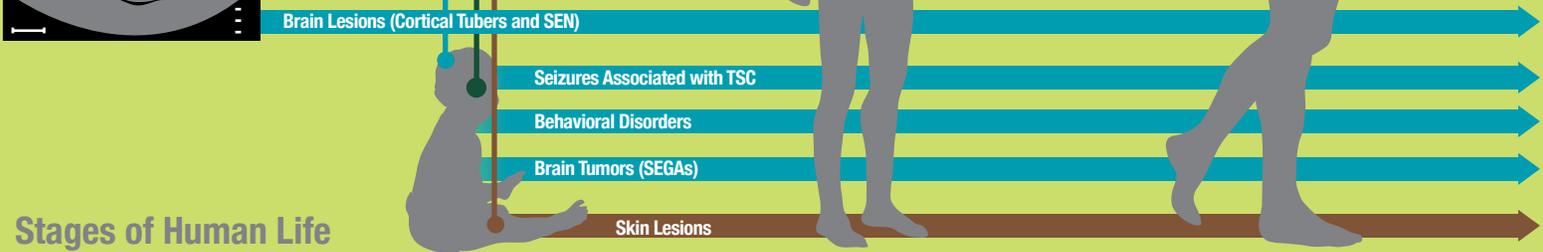
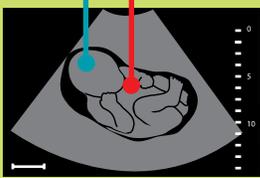
Brain Lesions

Subependymal Nodules (SEN)
 ~90% appearance rate in people with TSC
 Cortical Tubers
 >80% appearance rate in people with TSC



Heart Tumors

Cardiac Rhabdomyomas
 ~50% of newborns with TSC



Note: Manifestations are included above at the point during human development when the manifestation is known to first occur in a patient. Horizontal arrows indicate the stages of life during which a manifestation is known to occur. Additionally, the above statistics are taken from multiple studies and sources.