

Tuberous Sclerosis Complex (TSC)

➤ **What is Tuberous Sclerosis Complex (TSC) ?**

- Tuberous Sclerosis (also called *tuberous sclerosis complex*, or *TSC*) is a rare, multi-system genetic disease that causes non-cancerous (benign) tumors to grow in the brain and on other vital organs such as the kidneys, heart, eyes, lungs, and skin.

➤ **What are the signs of Tuberous Sclerosis Complex?**

- Tuberous sclerosis is often detected during infancy or childhood. Some people with tuberous sclerosis have such mild signs and symptoms that the condition isn't diagnosed until adulthood, or it goes undiagnosed. Others experience serious disabilities.
- Although the signs and symptoms are unique for each person with tuberous sclerosis, they can include:
 - **Skin abnormalities.** Most people with tuberous sclerosis have patches of light-colored skin, or they may develop small, harmless areas of thickened, smooth skin or reddish bumps under or around the nails. Facial growths that begin in childhood and resemble acne also are common.
 - **Seizures.** Growths in the brain may be associated with seizures, which can be the first symptom of tuberous sclerosis. In small children, a common type of seizure called infantile spasm shows up as repetitive spasms of the head and legs.
 - **Cognitive disabilities.** Tuberous sclerosis can be associated with developmental delays and sometimes intellectual disability or learning disabilities. Mental health disorders, such as autism spectrum disorder or attention-deficit/hyperactivity disorder (ADHD), also can occur.
 - **Behavioral problems.** Common behavioral problems may include hyperactivity, self-injury or aggression, or issues with social and emotional adjustment.
 - **Kidney problems.** Most people with tuberous sclerosis develop noncancerous growths on their kidneys, and they may develop more growths as they age.
 - **Heart issues.** Growths in the heart, if present, are usually largest at birth and shrink as the child gets older.

- **Lung problems.** Growths that develop in the lungs may cause coughing or shortness of breath, especially with physical activity or exercise. These benign lung tumors occur more often in women than in men.
- **Eye abnormalities.** Growths can appear as white patches on the light-sensitive tissue at the back of the eye (retina). These noncancerous growths don't always interfere with vision.

➤ **What is Tuberous Sclerosis Complex a result of?**

- *A random cell division error.* About two-thirds of people who have tuberous sclerosis have a new mutation in either the TSC1 or TSC2 gene—the genes associated with tuberous sclerosis—and do not have family history of tuberous sclerosis.
- *Inheritance.* About one-third of people who have tuberous sclerosis inherit an altered TSC1 or TSC2 gene from a parent who has the disorder.

➤ **What are some complications of TSC?**

- **Excess fluid in and around the brain.** One type of brain growth can block the flow of cerebral spinal fluid within the brain. This blockage can cause the buildup of fluid in the cavities (ventricles) deep within the brain, a condition called hydrocephalus. Various signs and symptoms include an unexpectedly large head size, nausea, headaches and behavior changes.
- **Heart complications.** Growths in the heart, usually in infants, can block blood flow or cause problems with heart rhythm (dysrhythmia).
- **Kidney damage.** Growths in the kidney can be large and cause potentially serious—even life-threatening—kidney problems. Growths in the kidney can cause high blood pressure or bleeding or lead to kidney failure. Rarely, kidney growths can become cancerous.
- **Lung failure.** Growths in the lungs can lead to a collapsed lung or fluid around the lungs that interferes with lung function.
- **Increased risk of cancerous (malignant) tumors.** Tuberous sclerosis is associated with an increased risk of developing malignant tumors in the kidneys and brain.
- **Vision damage.** Growths in the eye can interfere with vision if they block too much of the retina, though this is rare.

➤ Resources/References:

- <https://www.mayoclinic.org/diseases-conditions/tuberous-sclerosis/symptoms-cause/s/syc-20365969>
- <https://pulse.seattlechildrens.org/a-new-approach-tuberous-sclerosis-complex/>