



Understanding Tuberous Sclerosis Complex (TSC)

Tuberous Sclerosis Complex (TSC) is a **genetic condition** that causes **benign (non-cancerous) tumors** to grow in various parts of the body, most commonly the **brain, skin, kidneys, heart, and lungs**. It's not contagious and is caused by a change (mutation) in one of two genes, *TSC1* or *TSC2*. TSC affects people differently; some have very mild symptoms, while others have more significant health and developmental challenges. Early diagnosis and intervention are key to managing the condition effectively.

Key Medical Manifestations of TSC

TSC can affect many organ systems. The presence, number, and severity of these manifestations vary greatly among individuals.

Brain and Nervous System

- **Epilepsy/Seizures:** Often one of the first and most common signs. Seizures can range from mild to severe and may require medication to control.
- **Subependymal Giant Cell Astrocytomas (SEGAs):** Benign tumors in the brain's fluid-filled spaces that can sometimes grow and block the flow of fluid, requiring treatment.
- **Cortical Tubers:** Areas of abnormal brain tissue in the cortex (the outer layer of the brain).

Skin

- **Facial Angiofibromas:** Reddish spots or bumps, usually on the nose and cheeks, that may look like acne.
- **Shagreen Patches:** Thick, rough, or "orange peel" like patches of skin, often on the lower back.
- **Hypomelanotic Macules ("Ash-Leaf Spots"):** Small, lighter-than-normal spots of skin that are often leaf-shaped.
- **Ungual Fibromas:** Bumps that grow around or under the fingernails or toenails.

Kidneys

- **Angiomyolipomas (AMLs):** Benign tumors in the kidneys. These are very common and can sometimes cause problems like bleeding if they grow large.
- **Cysts:** Fluid-filled sacs in the kidneys.

Heart

- **Rhabdomyomas:** Benign tumors in the heart, typically seen in infants and young children, which often shrink on their own over time.

Lungs

- **Lymphangiomyomatosis (LAM):** Affects primarily adult women, causing cysts in the lungs that can lead to breathing problems.

Eyes

- **Retinal Hamartomas:** Benign tumors on the back of the eye (retina), usually don't affect vision.

TSC-Associated Neuropsychiatric Disorders (TAND)

TAND is a term used to describe the **brain-based behavioral, intellectual, and mental health challenges** that are very common in individuals with TSC. These issues stem from the changes in brain structure and function caused by TSC. Recognizing and addressing TAND is crucial for improving quality of life, educational outcomes, and social integration.

Domain	Examples of TAND Manifestations	Support and Awareness
Developmental	Intellectual Disability (learning challenges), Delayed Milestones (e.g., speech, motor skills).	Understand that learning may take longer and require specialized, consistent teaching methods. Focus on strengths.
Academic	Difficulties with Math, Reading, Writing, and Executive Function (planning, organizing, flexible thinking).	Provide structured routines, visual aids, and break down tasks into smaller, manageable steps.
Neuropsychological	Attention Deficit Hyperactivity Disorder (ADHD) (inattention, impulsivity, hyperactivity), Memory problems .	Offer frequent breaks, quiet workspace, and reminders. Use multi-sensory approaches to learning.
Psychiatric	Autism Spectrum Disorder (ASD) (social difficulties, repetitive behaviors), Anxiety, Depression, Aggression, Sleep Issues .	Look for triggers, teach coping strategies, provide a predictable environment, and consult a mental health professional specializing in TSC or related disorders.
Behavioral	Self-injury, Aggression, Tantrums .	Implement positive behavior supports and consistent discipline strategies. Communicate with the care team about changes.

