Tuberous Sclerosis Complex (TSC)
For Healthcare Providers

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools (www.gemssforschools.org).

Physical characteristics and/or symptoms

Note: not all people with TSC will have all of these features.

Common features of Tuberous Sclerosis Complex

Skin findings

- Facial angiofibromas.
- Hypomelanotic macules (ash-leaf spots)
- Shagreen patches
- Confetti skin lesions
- Fibrous cephalic plaques (raised lesions on forehead or scalp)
- Ungual fibromas

Central Nervous system

- Cortical brain tubers
- Subependymal nodules
- Subependymal giant cell astrocytomas (SEGA).
- Cortical brain tubers and subependymal nodules are thought to be associated with a higher risk for seizures, or issues with learning and behavior
- Seizures occur in 60-90% of people who have TSC.

Renal abnormalities

- About 80% of children with TSC have a renal lesion by 10.5 years.
- Angiomyolipomas
- Cysts
- Renal cell carcinomas

Cardiac abnormalities

- Rhabdomyomas.
- Arrhythmias

Pulmonary

- Lymphangioleimyomatosis (LAM)

- More common in females than males (40% of woman have LAM)

Eyes

- Multiple retinal nodular hamartomas
- Retinal achromic patch

Dental

- Intraoral hamartomas
- Dental pits

Developmental and cognitive

- Intellectual disability/developmental delay occurs in roughly 50% of individuals with TSC.
- TSC-associated neuropsychiatric disorder (TAND) - refers to the interrelated functional and clinical manifestations of brain dysfunction common in individuals with TSC, including behavioral, psychiatric, intellectual, academic, neuropsychological, and psychosocial difficulties Attention deficit hyperactivity disorder (ADHD)
- Behavioral and psychiatric disorders, often part of the autism spectrum disorders (ASD)

Recommended Routine Surveillance

- Brain MRI every 1-3 years in asymptomatic individuals less than 25 years to monitor for new occurrence of SEGAs
- Routine EEGs in individuals with known or suspected seizures
- Screening for TSC- associated neuropsychiatric disorders (TAND)
- Cardiac surveillance of cardiac rhabdomyomas
- Ophthalmologic evaluation as needed
- Monitor renal angiomyolipoma and renal cystic disease; asses renal function
- Clinical screening for LAM symptoms in woman older than age 18 years
- Annual dermatologic examinations
- Routine dental care

Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Emergencies should be handled as with any child.
- If seizures are present, the following seizure action plan may be useful: https://www.aap.org/en-us/Documents/Seizure_Action_Plan_for%20School.pdf

Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Cardiologist
    - Surveillance for cardiac rhabdomyomas
- Dermatologist
    - Hypomelanotic macules
    - Confetti skin lesions
    - Facial angiofibromas
    - Shagreen patches
    - Fibrous cephalic plaques
    - Ungual fibromas
- Developmental specialist
    - Speech therapy
    - Physical therapy
    - Occupational therapy
    - TSC-associated neuropsychiatric disorder (TAND)
    - Autism spectrum disorder
    - Attention deficit hyperactivity disorder
- Geneticist / Genetic Counselor
    - Diagnosis
    - Coordination of care
    - Genetic risk for family
    - Clinical trials
- Nephrologist:
    - Renal disease
- Neuroendocrinology
    - Neuroendocrine tumors
- Neurologist
    - Seizures
    - CNS tumors
- Ophthalmology surveillance:
    - Retinal lesions
- Psychiatrist/Psychologist
    - TSC-associated neuropsychiatric disorder (TAND)
- Pulmonologist
    - LAM evaluations

Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

My patient_____ has been diagnosed with Tuberous Sclerosis Complex (TSC). TSC is characterized by abnormalities of the skin, brain, kidneys, heart and lungs and

developmental delays. Medical complications with TSC syndrome include management of seizures, brain tumors, cardiac lesions, and kidney disease. Because of these, _____ needs the following accommodations.

Seven Important Aspects of School Life

"Tuberous Sclerosis Syndrome at a Glance" will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions

Resources

GeneReviews
https://www.ncbi.nlm.nih.gov/books/NBK1220/

Tuberous Sclerosis Alliance
http://www.tsalliance.org