TUBEROUS SCLEROSIS COMPLEX

What is it?

Tuberous Sclerosis Complex (TSC) is a multi-system genetic disease resulting from a mutation of the tumor suppressor gene. It leads to non-malignant growths in various organs of the body. These growths are sometimes referred to as tubers or lesions. Although tumors can grow anywhere, areas most commonly affected include the brain, eyes, heart, kidney, skin and lungs. Effects on the brain include seizures, developmental delay, intellectual disability and autism. Development and behavior can be greatly effected.

The growth of tumors resulting from TSC is not as severely unregulated as in cancer but these tumors may still cause serious problems. Such tumors can block the flow of cerebrospinal fluid (CSF) causing headaches, dizziness, changes in blood flow of the heart causing arrhythmia, blockage of the retina that affect vision, as well as disruptions of kidney function. There is no single clinical feature that is absolutely specific to TSC. Instead, a diagnosis of TSC is based on physical examination and imaging. A diagnosis usually includes at least 2 major TSC features.

TSC is caused by either an inherited gene from one parent (1/3 of cases) or the result of spontaneous mutation (2/3 of cases). However, many children are undiagnosed because of the unfamiliarity of TSC.

What are the characteristics or complications?

Tumors can form on many surfaces and layers of the brain. Lesions can form in the fluid filled brain ventricles and even potentially block the flow of CSF causing increased pressure. Some children may already have a shunt in place to drain increased fluid in the brain. Children experiencing headaches, nausea and vomiting, as well as changes in appetite and mood should be assessed by a neurologist.

Tumors can also affect the kidneys causing cysts and even possible bleeding. Tumors can also grow on cardiac tissue, lung tissue, as well as affect the retina of the eye. Most noticeable may be changes in skin growth and pigmentation. TSC patients may also experience seizures, developmental delays and a range of intellectual disabilities. Behavior problems can include aggression, self-harming behavior, specific phobias, sleep disturbances, and anxiety. There is also strong evidence of correlation for increased rates of ADHD and autism.

What is the treatment?

There is no cure for TSC but early diagnosis and intervention can greatly impact the person's development and quality of life. The goal of treatment is to provide the best possible quality of life with the fewest complications from the underlying disease process. Epilepsy is the main complication of TSC that requires long term medical treatment. Initially, a medication regime to control seizures is helpful but when its not, other treatments aimed at seizure control can be utilized. The Ketogenic diet may be used as a dietary treatment. Surgery is sometimes used when tumors that are blocking vital bodily function can be accessed and surgically removed. Drug treatment that can shrink tumors is also sometimes prescribed.

Kennedy Krieger Institute

The **Specialized Health Needs Interagency Collaboration (SHNIC)** program is a collaborative partnership between the Kennedy Krieger Institute and the Maryland State Department of Education.

Suggested school accommodations

Children with TSC can present differently in the educational environment. Although some children function within the normal IQ range, others may have some degree of intellectual disability. Common factors that have been identified include language and communication barriers, attention deficits, behavior outbursts, and social interaction disorders. Such barriers and their progression may be directly related to the severity and/or control of seizures. Supporting students with TSC in the school setting require educators and parents/guardians to work as a team. Some accommodations to consider for a 504/IEP could include:

- Neuropsychological assessment
- Behavior assessment
- PT/OT/SLP evaluations
- Vision consultation
- Maintain a quiet, calm environment
- Maintain structure, daily routines
- Consider extra adult support
- Extended processing time
- Preferential seating
- Offer clear, concise direction
- Repeat instructions
- Check for understanding
- Use step-by-step instructions
- Offer information in outline form
- Require organization, planning
- Limit "multi-tasking"
- Obsessive behaviors
- Need structure, routine
- Encourage peer groups
- Practice social skills through role-playing
- Note difficulty with fine and/or gross motor
- Consider adaptive or assistive technology
- Staff education/training as appropriate
- Emergency Evacuation Plan (EEP)

Specific health issues for Individualized Healthcare Plan

- Diagnosis including all medical characteristics
- Documentation of type of seizure, description of, typical length, characteristics, triggers, warning signs, how often seizures occur, and student's behavior following a seizure
- Baseline cardiac assessment (including blood pressure related to kidney function), lung assessment and skin assessment
- Orders for hidden devices such as a shunt or VNS, education about how to use/manage devices
- Current medication list including PRN medication
- Dose, route, time to administer emergency seizure medications
- Documentation/log of seizures
- Nutrition orders (Ex. Ketogenic diet)
- Safety precautions for ambulating, transitioning in hallways, wearing a helmet, etc.
- Return to class protocol
- Communicate with school staff, parents/guardian, and provider any changes or concerns about the disease
- Emergency Care Plan(s) (ECP) related to medical needs in the school setting and staff education/training as appropriate for each

Resources & Manuals

Kennedy Krieger Institute: Tuberous Sclerosis Clinic
https://www.kennedykrieger.org/patient-care/centers-and-programs/tuberous-sclerosis-clinic

National Association for Rare Disorders NORD: Tuberous Sclerosis
https://rarediseases.org/rare-diseases/tuberous-sclerosis/

Teacher's Guide: Educating a child with Tuberous Sclerosis Complex
http://citeseerx.ist.psu.edu/viewdoc/download?doi=10.1.1.666.378&rep=rep1&type=pdf

Tuberous Sclerosis Association
http://www.tuberous-sclerosis.org

Tuberous Sclerosis Alliance
http://www.tsalliance.org/pages.aspx?content=2