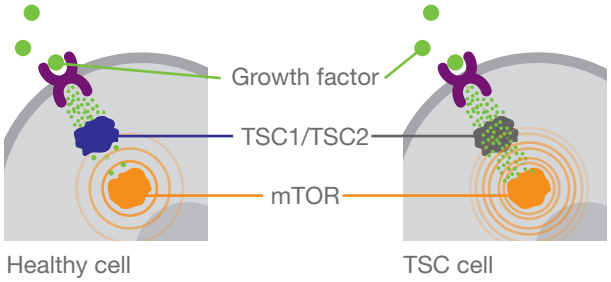
TSC Common Threads

Tuberous sclerosis complex affects each person differently and manifestations can change over time, but most share TSC genetic threads in common.

What is TSC?

The genetic threads that most people with TSC share in common are a defect in the TSC1 or TSC2 genes. Scientists believe that when these genes are defective, activity in a signaling pathway called mTOR (mammalian target of rapamycin), which acts as an important regulator of tumor cell division, blood vessel growth and cell metabolism, is deregulated. This can cause uncontrolled tumor cell growth and proliferation, blood vessel growth and altered cellular metabolism.



How is TSC Diagnosed?

Diagnostic tools such as CT scans and MRIs of the brain and ultrasounds of the heart, lung and kidneys may be used to help identify TSC features. There are two ways to make a definite TSC diagnosis:

1. **Genetic diagnostic criteria:** A mutation in either the TSC1 or TSC2 genes

2. **Clinical diagnostic criteria:** There are 11 major features and six minor features of TSC which span various manifestations. An individual must have two major features or one major feature with two or more minor features to be diagnosed definitively

Up to
1 MILLION
people worldwide have TSC

Approximately
50,000
people in the US have TSC

TSC occurs in all races and ethnic groups, and in both genders

The disease affects an estimated
1 in 6,000
newborns

Diseases with similar US prevalence rates include cystic fibrosis (approximately 30,000 people) and amyotrophic lateral sclerosis (ALS), or Lou Gehrig's disease (up to approximately 30,000 people)

About
1/3
of all people with TSC genetically inherit the disease, while in the remaining individuals, the disease is acquired as a result of spontaneous genetic mutation

TSC Signs, Symptoms and Resulting Disorders

Incidence in TSC Population

Signs, symptoms and resulting disorders can include:

Skin lesions >95%

Skin lesions can appear in many different forms, including reddish raised lesions or light, oval patches of skin

Seizures 85%

Seizures can start in infancy and may increase in frequency and severity throughout childhood

Kidney tumors ~60%

Kidney tumors (renal angiomyolipomas) can be difficult to manage and can be a major factor of morbidity and mortality in adult patients

Developmental disorders ~60%

Developmental disorders can range from mild learning disabilities to severe mental retardation

Heart tumors ~50%

Heart tumors (cardiac rhabdomyomas) frequently occur in infancy and are often used to help diagnose TSC

Autism ~50%

TSC is the leading known genetic cause of autism

LAM <40% ♀ & <12% ♂

Lymphangioleiomyomatosis (LAM) is a lung disease that can cause shortness of breath, occurring mostly in women and a small number of men with TSC

SEGAs ~20%

Subependymal giant cell astrocytomas (SEGAs) are non-cancerous brain tumors that may cause potentially life-threatening brain swelling if they grow

Note: not all people with TSC are affected by every TSC manifestation and resulting disorders vary from patient to patient

How is TSC Managed?

Monitoring/Screening: Because TSC is a lifelong condition, individuals need to be monitored by a doctor to ensure they are receiving the best possible care.

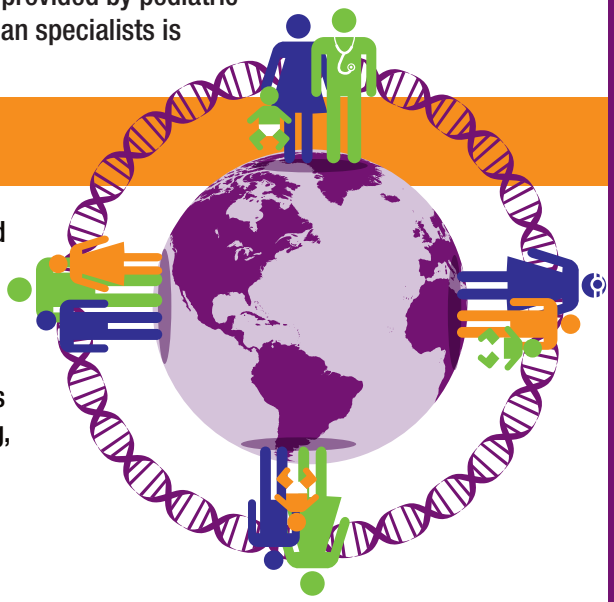
Specialized and Multidisciplinary Care: Due to the varied manifestations of TSC, care by a clinician experienced with the disorder is optimal. To address the multi-system nature of the disease, many people with TSC may see several doctors such as a neurologist, nephrologist, urologist, dermatologist, geneticist and pulmonologist. For example, specialty medical centers that have TSC clinics are set up with multidisciplinary care teams of physicians to help people with TSC address the various symptoms of the disease.

Transition Management: As people with TSC age out of pediatric management and enter adulthood, they may experience a gap in care. Patients who were treated by pediatric TSC specialists may switch to adult physicians specializing in their manifestations but who may not be equipped to offer the comprehensive care provided by pediatric specialists. This is why seeking care from a multi-disciplinary group of physician specialists is recommended for those with TSC.

TSC Community and Support

Connecting with other people living with TSC can teach those newly diagnosed and their families how to navigate the many domains of TSC, including doctors, hospitals, therapies, schools and services, as well as receiving and providing community and emotional support and perspective.

Meeting at local support groups and events, in addition to connecting online, is also helpful. To find support resources near you, visit www.tscinternational.org, or www.tsalliance.org in the US.



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