

# **Questions and Answers About Tuberous Sclerosis Complex**

## **Q: What is tuberous sclerosis complex (TSC)?**

A: Tuberous sclerosis complex (TSC) is a genetic condition characterized by lesions of multiple organs including the skin and central nervous system. Individuals with TSC often have growth of benign tumors and seizure disorders (epilepsy). The disease affects some people severely, while others are so mildly affected that they often remain undiagnosed. Some people with TSC experience developmental delay, mental retardation and autism. However, there are also many people with TSC that are living independent, healthy lives enjoying challenging professions such as doctors, lawyers, educators and researchers.

## **Q: How many people have TSC?**

A: At least two children born each day in the United States will have TSC. Current estimates of newborn babies affected with TSC are 1 in 6,000. Nearly one million people worldwide are known to have TSC, with approximately 50,000 in the United States. There are many undiagnosed cases of TSC due to the obscurity of the disease and the mild symptoms that occur in some people. TSC is as common as ALS (Lou Gehrig's Disease) or Duchenne's Muscular Dystrophy but is virtually unknown by the general population.

## **Q: How does a person develop TSC?**

A: TSC is transmitted either through genetic inheritance or as a spontaneous genetic mutation. Children have a 50% chance of inheriting TSC if one of their parents has this condition. At this point, only one-third of TSC cases are known to be inherited. The other two-thirds are believed to be the result of spontaneous mutation.

## **Q: If a parent has a mild form of TSC, will their child with TSC also be mildly affected?**

A: While this is possible, people with mild cases of TSC can produce a child who is much more severely affected. In fact, some people have such mild cases that they may only find out they also have tuberous sclerosis after their more severely affected child is diagnosed.

## **Q: How is TSC diagnosed?**

A: A clinical diagnosis of TSC is made after the following tests are performed: a brain MRI and/or CT Scan, renal ultrasound, echocardiogram of the heart, EKG, eye exam and a Wood's Lamp evaluation of the skin. These tests are able to detect almost all cases of TSC. In addition genetic testing for TSC is now available. However, while potentially providing a very clear diagnosis, genetic testing is not as sensitive as clinical-based testing and should always be undertaken in conjunction with genetic counseling.

## **Q: What genes are responsible for TSC?**

A: Two genes have been identified that cause TSC. Only one of the genes needs to be affected for TSC to be present. The *TSC1* gene is located on chromosome 9 and is also called the hamartin gene. The other gene, *TSC2*,



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is located on chromosome 16 and is called the tuberlin gene. Researchers are now trying to determine what these genes do and how defects in these genes cause TSC.

**Q: How can so many different organs be affected by TSC?**

A: Both the *TSC1* and *TSC2* genes are believed to control cell growth in the body. When either of these genes is defective, growth is not inhibited and TSC results. The genes also play a role in the early fetal development of the brain and skin.

**Q: Are the tumors cancerous?**

A: The tumors resulting from TSC are non-malignant, but may still cause problems. Tumors that grow in the brain can block the flow of cerebral spinal fluid in the spaces (ventricles) in the brain. This can lead to behavior changes, nausea, lethargy, headaches or a number of other symptoms. In the heart, the tumors are usually at their largest at birth, and then decrease in size as the individual gets older. These heart tumors, called cardiac rhabdomyomas, can cause problems at birth if they are blocking the flow of blood or causing severe arrhythmias. The tumors in the eyes are not as common, but can present problems if they grow and block too much of the retina. The tumors in the kidney (renal angiomyolipoma) can become so large they eventually take over normal function leading to kidney failure. Very rarely (less than 2%) individuals with tuberous sclerosis can develop malignant (cancerous) kidney tumors. In addition, there is a form of TSC that only affects the lungs of adult women. This disease process, called lymphangioleiomyomatosis (LAM) can also exhibit aggressive behavior but fortunately is rare.

**Q: What is the normal life expectancy of an individual with TSC?**

A: Most people with TSC will live a normal life span. There can be complications in some organs such as the kidneys and brain that can lead to severe difficulties and even death if left untreated. To reduce these dangers, people with TSC should be monitored throughout their life by experienced physicians for potential complications. Thanks to research advancements and improved medical therapies, people with TSC can expect improved health care and quality of life.

**Q: Since there is no cure, what can be done?**

A: Early intervention is key. Advancements in research are bringing new and improved therapeutic options. Surgery to remove tumors or stop tumor growth is helping to preserve the function of affected organs. New technology and research is pinpointing the exact location of the brain initiating seizures and creating new therapies to help control epilepsy. With every new day we are one step closer to better and better treatments.

You can stay informed of the advancements that are made by becoming a Tuberous Sclerosis Alliance member and receiving our quarterly magazine , *Perspective*. For the latest information, visit [www.tsalliance.org](http://www.tsalliance.org).

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*\*\*Tuberous Sclerosis Alliance Information Sheets are intended to provide basic information about TS. They are not intended to, nor do they, constitute medical or other advice. Readers are warned not to take any action with regard to medical treatment without first consulting a physician. The TS Alliance does not promote or recommend any treatment, therapy, institution or health care plan.*

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