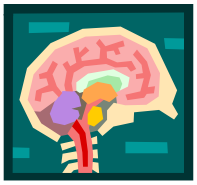

Disability Awareness Begins With You: Tuberous Sclerosis Complex

What is tuberous sclerosis complex (TSC)?
Tuberous sclerosis complex is a genetic condition characterized by lesions of the skin and central nervous system, tumor growth and seizures. The disease affects some people severely, while others are so mildly affected that it often goes undiagnosed. Some people with tuberous sclerosis experience developmental delay, mental retardation and autism. However, there are also many people with tuberous sclerosis living independent, healthy lives who are enjoying challenging professions such as doctors, lawyers, educators and researchers.

How many people have tuberous sclerosis?
At least two children born each day will have tuberous sclerosis. Current estimates place tuberous sclerosis affected births at one in 6,000. Nearly 1 million people worldwide are known to have tuberous sclerosis, with approximately 50,000 in the United States. There are many undiagnosed cases due to the obscurity of the disease and the mild form symptoms may take in some people. TSC is as common as ALS (Lou Gehrig's Disease) but virtually unknown by the general population.

How does a person develop tuberous sclerosis?
Tuberous sclerosis is transmitted either through genetic inheritance or as a spontaneous genetic mutation. Children have a 50 percent 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 a result of spontaneous mutation. The cause of these mutations is still a mystery.

If a parent has a mild form of tuberous sclerosis, will their child with tuberous sclerosis also be mildly affected?
People with mild cases of tuberous sclerosis can produce a child who is more severely affected. In fact, some people are so mildly affected that they may only find out they also have TSC after their more severely affected child receives a diagnosis of TSC.

How is tuberous sclerosis diagnosed?
Diagnosis of tuberous sclerosis is currently made after the following tests are preformed: a brain MRI or CT Scan, renal ultrasound, echocardiogram of the heart, EKG, eye exam and a Wood's Lamp evaluation of the skin.

What genes are responsible for tuberous sclerosis?
Two genes have been identified that can cause tuberous sclerosis. Only one of the genes needs to be affected for tuberous sclerosis to be present. The TSC1 gene is located on chromosome 9 and is called the hamartin gene. The other gene, TSC2, is located on chromosome 16 and is called the tuberin gene. Researchers are now trying to determine what these genes do and how a defect in these genes causes tuberous sclerosis.

How can so many different organs be affected by tuberous sclerosis?
Both the TSC1 and TSC2 genes are believed to suppress tumor growth in the body. When either of these genes are defective, tumors are not suppressed and tuberous sclerosis results. The genes also play a role in the early fetal development of the brain and skin.

Are the tumors cancerous?
The tumors resulting from tuberous sclerosis are non-cancerous, 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, 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 arrhythmia problems. 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 all of the normal kidney function. In the past, the patient was left until they developed kidney failure. Today, doctors are more aggressive and remove individual tumors before they get too large and compromise healthy kidney tissue. Very rarely (less than 2 percent of) individuals with TSC develop malignant (cancerous) kidney tumors.

What is the normal life expectancy of an individual with tuberous sclerosis?
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 their physician for potential complications. Thanks to research findings and improved medical therapies, people with tuberous sclerosis can expect improved health care.

Since there is no cure, what can be done?
Early intervention is helping to overcome developmental delays. 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. Technology is pinpointing the exact portions of the brain stimulating seizures and creating new therapies to help control seizures. With every new day we are one step closer to finding improved treatments.

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http://www.tsalliance.org/pages.aspx?content=117