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Dear Family Member:

I'm writing because a member of your family has been diagnosed with tuberous sclerosis complex, or TSC.

TSC affects everyone differently, which makes it challenging to understand the disease and to know what to be concerned about and what not to worry about. TSC affects some people severely, while others are so mildly affected, they often remain undiagnosed. Some people with TSC may experience developmental delays and intellectual disability. However, many people with TSC live independent, healthy lives enjoying challenging professions such as doctors, lawyers, educators and researchers.

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 1 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 amyotrophic lateral sclerosis (Lou Gehrig's Disease) or Duchene's muscular dystrophy but is virtually unknown by the general population.

TSC is caused by mutations in one of two genes: TSC1 or TSC2. Genetic testing is available that can identify the mutation that causes the disease in 85 - 90% of people with TSC. This can be useful for confirming diagnosis and for family planning purposes. If a causative mutation is found in you or a family member, you should consider talking with your doctor and/or meeting with a genetic counselor to discuss these issues further.

TSC is transmitted either through genetic inheritance or as a spontaneous genetic mutation. Two-thirds of TSC cases are the result of spontaneous mutation, meaning neither parent carries a mutation. However, one-third of TSC cases are inherited from a parent. Children have a 50% chance of inheriting TSC if one of their parents has this condition. A parent with a mild case of TSC can produce a child who is more severely affected. In fact, some people have such mild cases they may only find out they also have TSC after their more severely affected child is diagnosed.

TSC can cause the growth of non-malignant tumors in various organs and leads to an increased risk of epilepsy, autism, and cognitive or developmental delay. However, most people with TSC have many, but not all, of the possible manifestations. Family members of infants diagnosed with TSC should particularly watch for signs of infantile spasms, a

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particularly dangerous form of epilepsy that leads to cognitive impairment and delays intellectual and motor development.

The tumors resulting from TSC are non-malignant but may still cause problems. Tumors that grow in the brain are generally diagnosed in infancy or childhood and can block the flow of fluid in the brain. Heart tumors, called cardiac rhabdomyomas, may be found by ultrasound during pregnancy or soon after birth, and they usually shrink during infancy. Tumors in the kidney (called renal angiomyolipomas) are generally diagnosed in adulthood and can lead to bleeding or loss of kidney function.

In addition, women with TSC are at risk of developing a lung condition known as lymphangioleiomyomatosis (LAM), although LAM can rarely occur in men, also. Tumors on the skin, known as angiofibromas, often appear during childhood and adolescence and can be disfiguring and bleed easily if scratched.

Fortunately, treatment options for TSC have expanded greatly in the last decade. Afinitor (everolimus)® is a drug that shrinks and stops the growth of tumors and is now FDA-approved for the treatment of subependymal giant cell astrocytomas (SEGAs) in the brain, angiomyolipomas in the kidney and as an added treatment to other antiseizures medications for some individuals with TSC. However, in some cases surgery may be required to remove particularly threatening tumors. Sabril (vigabatrin) ® and Acthar® Gel are approved for the treatment of infantile spasms. Epidiolex (cannabidiol) is approved of seizures associated with TSC. Additionally, we are learning more each day about better ways to treat epilepsy, angiomyolipomas, angiofibromas, and other aspects of TSC.

If you would like to connect with a doctor or TSC Clinic in your area, do not hesitate to contact the TSC Alliance. The TSC Alliance is the only national voluntary health organization dedicated to finding a cure for TSC while improving the lives of those affected. Much more information can be found on our website at tscalliance.org.

I hope this information is helpful to you. Should you have any questions or would like to get more information, please contact the TSC Alliance at (800) 225-6872.

Sincerely,

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