

MY CHILD HAS

# TUBEROUS SCLEROSIS

A BROCHURE  
FOR PARENTS

by The Tuberosus  
Sclerosis Alliance

The purpose of this brochure is to explain tuberous sclerosis and its variable features to parents; to outline some of the commonly needed medical tests and their meaning; and to help parents cope with tuberous sclerosis. The information here is an introductory overview. You should discuss specific problems in greater detail with your child's physicians.

First and foremost, you need to know that you are not the only parents to go through this. It is normal to ask "What is tuberous sclerosis?" and "Why is my child affected?"

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### Christine

Christine, 18 years old, was diagnosed with tuberous sclerosis (TS) at 6 months. At one point, she was having 50 to 80 infantile spasms daily. Fortunately, medications almost eliminated these seizures. During her school years, she experienced some very mild learning problems, but she still did well in the regular education system. Today, Christine enjoys horseback riding and volunteer work, and she looks forward to attending a community college.

### Jennifer

A typical 2-year-old, Jennifer suddenly began having seizures and was diagnosed with tuberous sclerosis. Her parents noticed that her language and social development slowed dramatically. For the next 10 years, Jennifer received intensive language therapy and special education instruction. Now, at age 12, she still experiences three to four seizures per week. Although several years behind her peers academically and socially, Jennifer is a happy, well-liked girl who is involved in Girl Scouts, is an excellent swimmer, and plays the piano.

### Robbie

Robbie, 7, was not diagnosed until a year ago when he visited a dermatologist because of red bumps forming across his cheeks and nose. These skin symptoms, along with the appearance of numerous white spots on his back and legs, confirmed the diagnosis of TS. Robbie's level of development is age-appropriate, and he is a typical little boy who loves to ride his bike and collect baseball cards.



*TS is a genetic disorder that causes tumors to form in many different organs*

### Jason

Jason, 23, had many seizures as an infant, but they stopped by age 2. His parents, however, noticed that he was very withdrawn. He seemed to be in “his own little world” and did not speak. Over the years, Jason became increasingly aggressive and difficult for his parents to handle. At age 12, he was placed in a small group home. Jason comes home to his parents on weekends, and they report that in the past few years his behavior has improved and that he now enjoys social activities.

### Michael

Michael, 14, appears to be a typical teenager. He likes listening to music, playing the guitar, and skateboarding. He is very self-conscious, though, about the abundance of small red tumors on his face caused by tuberous sclerosis. Because Michael is academically and musically talented, he tends to spend most of his time studying, reading, or playing the guitar. He is a loner and finds it difficult to make friends. Michael has just begun counseling in hopes of gaining some social skills. He also is considering having laser surgery on his face.

### Susan

Susan, 35, is a married, college-educated librarian. She was diagnosed with tuberous sclerosis as a baby. She had mild seizures as a child but has not taken anticonvulsants since age 12. Susan looks back on her childhood as happy and normal. Her only symptom during adulthood has been the formation of small tumors on her kidneys and lung cysts. These tumors have not yet caused any major medical problems.

The fact that all of these people have tuberous sclerosis shows how the disease can affect people in very different ways.

### **Tuberous Sclerosis—What Is It?**

Tuberous Sclerosis is a genetic disorder that causes tumors to form in many different organs—primarily in the brain, eyes, heart, kidney, skin, and lungs. You will see it referred to both as tuberous sclerosis (TS) and tuberous sclerosis complex (TSC). (The term TSC is used in the scientific literature to distinguish tuberous sclerosis from Tourette’s syndrome.) The true prevalence of TS is unknown, but its incidence has recently been estimated to be 1 in 6,000 live births. This means approximately 50,000 individuals in the United States and more than 1,000,000 worldwide have TS. It occurs in both sexes and in all races and ethnic groups.

Because TS is a genetic disease, it is not contagious (you cannot catch it from other people). It results from a genetic mutation (change) over which a parent has no control. The genetic nature and inheritance of TS will be further discussed in the section titled Genetic Counseling. It is often first recognized in children because of two neurologic symptoms—epileptic seizures and/or varying degrees of mental handicap. However, the clinical symptoms of TS vary greatly and may not appear until later in life (see the section titled Physical and Mental Symptoms). We cannot presently predict who will develop a particular symptom and who will not.

### **Physical and Mental Symptoms**

TS in an infant is usually suspected when the child has cardiac rhabdomyomas (benign heart tumors) at birth or begins to have seizures, especially infantile spasms. These symptoms combined with a careful examination of the skin and brain make it possible to diagnose TS in a very young infant. However, many children are not diagnosed until later in life when they begin to show symptoms such as seizures, a reddish rash on their face (facial angiofibromas), or developmental delays with or without



seizures or other signs of TS. The skin signs of TS assist in the diagnosis but usually do not cause severe problems for the child. The text that follows describes the various symptoms of TS, as well as the tests that should be used to help physicians determine if your child has tuberous sclerosis. Don't be afraid to ask your child's physicians any questions you have. It often helps to write down your questions at home and bring them with you to your appointment with the physician.

**Skin Lesions (Markings or Growths).** An examination of your child's skin is relatively simple and can provide important clues in the diagnosis of TS. Here is what the physician will be looking for:

*The skin lesions that are most often treated are the facial angiofibromas and the peri- and subungual fibromas*

- **Hypomelanotic macules.** The physician may do a careful examination of the skin using a Wood's lamp (a special ultra-violet [UV] light) that can make hypomelanotic macules (white spots) more obvious. When the skin is illuminated with the UV lamp, these macules appear very different from the surrounding skin. These white spots may appear on any part of the body and can be of any shape. Some may have an oval shape, while others are elongated and resemble the leaf of an ash tree ("ash-leaf spot"). The white spots can look like speckling, especially on the legs and arms (sometimes referred to as "confetti").
- **Facial angiofibromas** are small reddish spots or bumps that typically appear across the cheeks and nose. They may be present at birth but more often appear as the child reaches 4 or 5 years of age. As the child grows older, these angiofibromas may spread around the nose and lower lip and sometimes onto the chin and become larger. Angiofibromas are actually small, slow-growing, benign tumors grouped together.

- **Other less common TS skin lesions** are the **shagreen patch**, a patch of thickened and elevated pebbly skin (like an orange peel) usually found on the lower back and nape of the neck, although sometimes seen on other parts of the body; **peri- or subungual fibromas**, small, wart-like tumors that develop around or under the fingernails and toenails, which are usually not seen until later in life; and the **forehead plaque**, which is similar to the shagreen patch but is found on the forehead or scalp. Again, these skin lesions can be seen and felt with examination of the skin.

The skin lesions that are most often treated are the facial angiofibromas and the peri- and subungual fibromas. Facial angiofibromas can be removed using dermabrasion or laser treatment; because the angiofibromas are caused by a genetic condition, these procedures should not be considered cosmetic surgery. Current research suggests that better results may be achieved if the angiofibromas are treated earlier rather than later. You should obtain a referral to either a dermatologist or a plastic surgeon for treatment of angiofibromas or unguinal fibromas.

**Brain and Neurologic Function.** Several types of brain lesions are seen in individuals with tuberous sclerosis; some people will have all of these lesions, whereas others will have no brain involvement at all.

- **Cortical tubers** (from which TS is named) can be thought of as a “birth defect” on the brain. They are small areas in the cortex (the outer layer of the brain) that do not develop normally. It is thought that the presence of cortical tubers, which disrupts the normal “wiring” of the brain, is what causes seizures in individuals with TS.



- **Subependymal nodules** develop near the walls of the cerebral ventricles (the cavities in the brain that contain cerebrospinal fluid). Typically, these nodules accumulate calcium within the first few months or years of life. Because of this calcification, they can be easily detected with a computed tomography (CT) scan. The subependymal nodules are not directly responsible for neurological problems.
- **Subependymal giant cell astrocytomas (SEGAs)**. This type of benign tumor develops in approximately 15% of individuals with tuberous sclerosis. Typically, SEGAs do not occur in very young children, and the chance for their growth decreases after age 20. If a giant cell astrocytoma grows large enough, it can block the flow of fluid inside the ventricles of the brain, and the tumor will have to be removed and/or the ventricles shunted to relieve fluid buildup and pressure. Symptoms include vomiting, nausea, and headaches, as well as changes in appetite, behavior, and mood. These symptoms may or may not signal growth of a tumor, but they do signify that there may be a problem and that the child should be seen by a physician. Brain imaging should be done at the time of diagnosis to get a baseline image and then every 1 to 3 years afterwards. A brain scan examination can sometimes show growth of a tumor even before symptoms develop.
- **Epilepsy/seizure disorders**. Seizures occur in 60% to 90% of individuals with tuberous sclerosis. They are often the first symptom of TS. Watching a child have a seizure is a frightening experience for parents. Early in life, the seizures may consist only of brief head nodding or staring, or they may be more generalized, with spasms of the limbs and flexion and extension of the head and trunk. In older children and young adults, the seizures sometimes occur less often or cease.

You should contact your physician at the onset of seizures or if the seizures become more frequent, prolonged, or change in manifestation. Your doctor will want to know what the child was doing right before the seizure, exactly what the child did during the seizure, how long it lasted, and how the child felt and acted afterward. It is difficult to remember all this, so write it down. You might be able to use a home video camera to videotape your child having a seizure. This can be valuable in showing your doctor what your child does during a seizure. If seizures are suspected, your child will probably have an **electroencephalogram (EEG)**. This will involve gluing many electrodes and wires to your child's scalp to record the electrical patterns generated by the brain. An EEG is not painful, but your child may need to be sedated so that he or she will lie still and not pull off the wires. Neurologists can detect abnormalities in the brain signals and determine if seizures are occurring and what type they are. It may be necessary to do a Video-EEG, which involves the same EEG recording plus videotaping your child over a period of hours or days so that the physicians can see what the seizures are like.

Seizure control in a child with TS is important. There appears to be a close relationship among the age of the child at the onset of seizures; their frequency, duration, and severity; and the degree of mental handicap. **Antiepileptic drugs (AEDs)**, properly administered, may control the seizures completely, although in some people the seizures return or cannot be controlled. The optimal dosage is the amount and combination of medication necessary to control the seizures without causing adverse side effects. Finding the optimal dosage is a real challenge for the physician and is, to some degree, a process of trial and error. Any medication can have adverse side effects even if most patients tolerate it well. You and your physician should discuss the possible side effects of



*It is important that your child receive education and therapy to develop to his or her full potential*

any AED your child takes. Also, because several different AEDs are available, your physician may try different drugs until optimal seizure control is reached.

Some children with TS have “infantile spasms.” During this type of seizure, the child brings his legs up to his chest in a repeated motion. Infantile spasms can sometimes be eliminated by the use of **adrenocorticotrophic hormone (ACTH)**, a hormone from the pituitary gland that stimulates the adrenal glands to produce cortisone. This medication is not used for long periods of time because of adverse side effects. Valproic acid (Depakene) has also been used to treat infantile spasms. Although it is not approved by the Food and Drug Administration (FDA), vigabatrin (Sabril) is used in many countries and is considered the most effective drug for the treatment of TS-related infantile spasms. You should discuss the use of these drugs with your neurologist.

You can get information about drugs and their side effects from your pharmacist when a drug is prescribed for your child. You can get more information on antiepileptic drugs from a copy of the *Physicians Desk Reference* at your public library, from the Tuberous Sclerosis Alliance, or from the Epilepsy Foundation of America (4351 Garden City Drive, Landover, MD 20785; 1-800-332-1000). Another resource is the Epilepsy Information Service, a 24-hour hotline for questions about seizures and antiepileptic drugs; the number to call is 1-800-642-0500.

**Mental Disability.** You may be wondering if your child will be mentally disabled and this is a difficult question. One-half to two-thirds of individuals with TS are mildly learning disabled to severely mentally retarded, but the percentage is dropping as more and more individuals with no mental handicap are being

diagnosed with TS. Some individuals with TS are neither mentally handicapped nor have epilepsy. Others have epilepsy but are not mentally handicapped. However, most individuals with TS who are mentally handicapped experienced seizures at some point in their early life.

Some children with TS who have mild mental disabilities can be mainstreamed in their schools and will develop both mental and motor skills similar to those of their peers. Others will attain a certain mental and/or motor skill level and then not progress beyond that point. It is extremely difficult to predict how your child will develop, but it is important that your child receive all of the education and therapy available to develop to his or her full potential. A few children with TS will regress to a lower level of achievement. This may be caused by a loss of seizure control, a severe seizure or seizures, growth of a brain tumor, or side effects from antiepileptic drugs. Regression may be temporary, especially if the cause can be detected and successfully treated.

**Psychiatric and Behavioral Problems.** The behavior of a child with TS can be the most difficult and trying problem for the parents and family. Aggression, sudden rage, hyperactivity, attention deficit, acting out, obsessive-compulsive behavior, repetitive behaviors, staying in their “own world,” being nonverbal even at an age when most children are speaking, and other autistic behaviors have all occurred in children with TS. Such behavior does not mean you are a bad parent; it is due to TS. Some children with TS, usually those who have a mental disability, are also diagnosed with autism. There appears to be a connection between TS and autism that is not understood, and active research is exploring this link. It is important for you to keep notes on your child’s behavior and on whether or not he or she reaches the developmental milestones, and bring them to the attention of your child’s physicians. The earlier



these behaviors are identified and special programs outlined for your child, the better.

Occasionally, individuals with TS are also diagnosed with schizophrenia, bipolar disease (manic depression), depression, or other psychiatric disorders. Again, bring any unusual, disruptive behaviors to the attention of your child's physicians and be your child's advocate so that he or she can obtain optimal medical and psychiatric treatment.

**Involvement of Organ Systems.** TS can affect various organs of the body. The major effects are discussed below.

- **Heart.** One or more cardiac rhabdomyomas (benign tumors) can form in the heart of an infant with TS. If these tumors are multiple and extensive, they can cause blockage and may even cause the death of a newborn. However, these tumors do not usually grow after birth and, in fact, usually decrease in size. If these tumors do not cause problems at birth, they may not be symptomatic during your child's life. An electrocardiogram (EKG) should be done to determine if conduction of the impulses in the walls of the heart is dysfunctional (the physician will be looking for conduction blocks or arrhythmias). If your child has rhabdomyomas, you should consult with your cardiologist before beginning treatment with ACTH, as this drug may cause the rhabdomyomas to grow.
- **Kidneys.** Renal angiomyolipomas (benign tumors) and cyst growth in the kidneys of individuals with TS can be serious. However, significant complications do not often develop before the second or third decade of life. At diagnosis, the child should have a baseline ultrasound, CT, or MRI examination of the kidneys so that any later tumor growth can be seen in comparison with this baseline scan. A scan should be

repeated at least every 1 to 3 years as long as the child shows no symptoms of kidney involvement. Angiomyolipomas larger than 4 cm are more likely to cause symptoms but may be treatable through surgical removal, partial embolization (blocking blood supply to the tumor), or other means. If they are smaller than 4 cm, they should be closely monitored. Blood pressure should be monitored at each visit to the physician because it can be the first sign of increasing kidney involvement. Other signs to watch for are blood in the urine and complaints of abdominal or low back pain. An individual with TS who has kidney involvement can go a very long time before the kidney becomes nonfunctional. Adults with TS have been successfully treated with both kidney dialysis and kidney transplantation. Although most tumors in TS are benign, there are some cases of renal cell carcinoma, which are malignant (dangerous) tumors on the kidneys.

- **Lungs.** Lung involvement occurs almost exclusively in women, and the average age of onset is during the childbearing years, suggesting that the disease could be estrogen related. However, a small number of men with lung disease have been reported. Many individuals who have lung involvement due to TS also have lymphangioleiomyomatosis (LAM), a degenerative cystic disease of the lungs. Female TS patients should have a high-resolution CT scan of the lungs performed at around age 18 to provide a baseline reference.
- **Eyes.** Benign tumors and depigmented patches may occur inside the eyes of individuals with TS, but they rarely cause any visual loss or problems. However, the presence of these tumors can assist in the diagnosis of TS. An ophthalmologist who is familiar with TS tumors of the eye should examine your child using indirect ophthalmoscopy with the pupils



dilated. The child may have to be sedated to improve cooperation during the exam.

- **Other Organ Systems.** Cysts and tumors similar to those observed in the kidney are often found in the liver, lung, pancreas, and other organs. These lesions are not usually seen until later in life and are rarely symptomatic. Bone cysts can also develop but usually do not cause problems until later in life. In addition, pits have been noted in both baby and adult teeth in over 90% of individuals with TS, but their significance is not known. Rectal polyps have also been reported but do not appear to cause problems.

**Vaccines.** The pertussis (P) vaccine is usually given to infants and young children in combination with the diphtheria (D) and tetanus (T) vaccines as the DPT vaccine. An association between the pertussis vaccine and acute neurologic problems has been suggested in children in general, and some children with TS have had their first seizures after receiving the pertussis vaccine. It is not clear if this first seizure was due to the pertussis vaccine, the fever that developed after the vaccination, or other causes. Some physicians believe that the seizures would have eventually started in any case, and many questions are unanswered about the link between the pertussis vaccine and seizures. Parents of a child with TS should approve administration of the pertussis vaccine only under the guidance of a pediatric neurologist. Several factors determine whether the pertussis vaccine should be given to your child, including the prevalence of whooping cough in your area. A new pertussis vaccine, called A-Cellular, may prove to produce fewer side effects and could be used in place of the standard pertussis vaccine. Remember that it is important to vaccinate your child with the measles, mumps, and rubella (MMR) and DT vac-

cines, even if you and your child's physician decide not to use the pertussis vaccine.

### **Diagnostic Screening and Follow-Up**

To summarize what was outlined in the previous section, the following tests are recommended at diagnosis and for follow-up screening:

**All of the following tests are recommended at the time of diagnosis of TS or if TS is suspected:**

1. A thorough examination of the skin, including an examination with a Wood's lamp, can detect the common skin lesions of TS.
2. Dilation of the pupils with eye drops containing an atropine-like substance allows direct examination of the retina with an ophthalmoscope. An ophthalmologist familiar with TS can also do an indirect examination with a magnifying lens.
3. A CT or MRI of the brain should identify TS brain lesions. A brain MRI examination better defines the extent of brain involvement in TS.
4. An ultrasound, CT, or MRI of the kidney should be done to document any kidney involvement.
5. An EEG should be done if the child has seizures.
6. An echocardiogram (to detect cardiac rhabdomyomas) and an EKG of the heart will document any heart involvement.



*A gene that carries a mutation can disrupt normal development and functioning*

You and your child's doctors should determine a follow-up screening schedule according to your child's health status. At the least, a yearly physical should be done and blood pressure should be monitored at every office visit, especially if there is kidney involvement. Your doctor will probably recommend periodic brain and kidney scans. In addition, screening for behavioral or neurologic problems will likely be ordered when the diagnosis of TS is made. Genetic testing for a mutation in one of the TSC genes (see the following section on Genetic Counseling) may be ordered. Of course, you should notify your child's physicians as soon as possible if your child's appetite, vision, or behavior changes suddenly or drastically. For additional information on diagnostic criteria and follow-up testing, please see referenced papers at the end of this brochure.

### **Genetic Counseling**

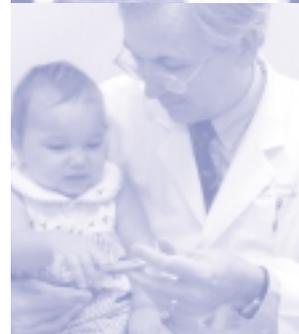
Genetic counseling is the process of giving patients and their families information about the genetic nature of their condition. The goal of genetic counseling is to ensure that the family understands the genetic implications of the diagnosis and to help the family make informed medical and personal decisions. Genetic counseling for TS often entails a discussion of the inheritance of TS, the chance that other family members may have undiagnosed TS, the possibility of TS occurring in future offspring, genetic (or DNA) testing for the causative mutation, available reproductive options, and issues surrounding acceptance of and adaptation to the diagnosis.

Tuberous sclerosis is a genetic condition, meaning that it is caused by a change, or mutation, in one gene. Genes are the instructions for the normal growth and maintenance of our bodies. Each of us has thousands of genes, and each gene is responsible for the direction of a specific protein or component of our bodies. A gene that carries a mutation is unable to

instruct the body to grow correctly, causing a disruption in normal development and functioning. All genes, including those involved in TS, come in pairs, with one copy inherited from (or passed down by) the mother, and the other copy inherited from the father. In some genetic conditions, like TS, a mutation in one copy of the gene is enough to cause the condition. These conditions are called dominant conditions because the mutation in one copy “dominates” over the normal copy, causing the condition and its symptoms. In other genetic conditions, both copies of the gene must have mutations before the symptoms occur. These are known as recessive conditions. One well-known recessive condition is cystic fibrosis.

Approximately 33%, or one-third, of individuals with TS inherit it from a parent who also has TS. When you have children, you pass on one copy of each of your gene pairs to the child, and your partner passes on one copy. If a parent has TS and passes on the copy of the gene with the mutation, then the child will also have TS. If the parent passes on the copy of the gene without the mutation, the child will not have TS. Thus, there is a 50% chance with each pregnancy for a parent with TS to have a child with TS. This is true regardless of the sex of the parent or the sex of the child.

In the remaining 66% or two-thirds of individuals with TS, neither parent shows any symptoms or signs of TS. It appears that one of the normal genes from one parent changes to the abnormal form, leading to a new (or sporadic) occurrence of TS in the child. Normally, these parents do not have another child with TS because the mutation was sporadic, not inherited. However, some families have more than one child with TS, even though neither parent showed symptoms or findings of TS. How does this occur?



*There is a 50% chance with each pregnancy for a parent with TS to have a child with TS*

Scientists have determined that a small number of *physically unaffected* parents of a child with TS actually have TS mutations in some of their cells. Because the mutation is limited to a small portion of all of the body's cells, these individuals show no signs of TS. But if a portion of the egg or sperm cells of a parent carries the TS mutation, that parent can have more than one affected child, possibly at the same 50/50 chance that persons with TS have. A person who carries cells with TS mutations in her egg or his sperm supply has germline mosaicism. "Mosaicism" means that the person's body is made up of a combination of cells with and cells without a TS mutation, and "germline" refers to the presence of cells with TS mutations in the egg or sperm cell supply. Germline mosaicism is relatively rare, and this explanation does not apply to most families with a sporadically affected child. However, the occurrence of germline mosaicism has led geneticists to estimate a recurrence risk (or chance that a family with a sporadically affected child will have another child with TS) ranging from 1% to 3%. At this time, there is no simple way to determine whether an unaffected parent of a child with TS has germline mosaicism.

Today, families who want to know what mutation caused TS in their child or other family member can pursue DNA testing of the two known TS genes. These genes are referred to as TSC1 and TSC2. With today's testing methods, 70% of the mutations can be identified. Because people can have TS and not have an identifiable mutation, the DNA test for TS cannot always be diagnostic, meaning if the mutation is not found, the test cannot be used to make the diagnosis in a person with questionable physical findings. However, it can be used to test other family members with questionable symptoms of TS or for prenatal diagnosis in persons who have TS or who are concerned about recurrence due to germline mosaicism. Such testing can be per-

formed only after the mutation has been found in the family member with a definite diagnosis of TS.

Despite advancing knowledge about TS mutations, we still cannot predict the severity of physical symptoms in a person with a new diagnosis of TS. A person can have TS and have very few or mild symptoms, while a family member with TS can have more severe or extensive symptoms. It is thought, however, that most people who have a TS mutation (excluding persons with germline mosaicism) will have some signs or symptoms if examined carefully by a physician familiar with the diagnosis of TS. The distinction between sporadic TS and familial (or inherited) TS is important, as it affects the chance for other persons in the family to be affected. Therefore, immediate family members of a person newly diagnosed with TS should be thoroughly examined.

These tests are recommended in the evaluation of parents and siblings of individuals with TS:

1. A family pedigree (or documentation of your family's medical history) should be developed that shows all of the members of your extended family and lists those who have had seizures, white spots, or any other sign of TS. This will greatly assist the geneticist in determining which other family members should be examined for a diagnosis of TS.
2. A thorough examination of the skin (usually including a Wood's light) should be done by a physician who is familiar with the skin abnormalities of TS.
3. An examination of the eyes should be done by an ophthalmologist who is familiar with TS to determine if any findings of TS are present.
4. A CT or MRI of the brain should demonstrate TS brain lesions.
5. Ultrasound, CT, or MRI of the kidneys may show kidney involvement.



*A positive attitude will help your child to achieve all that he or she can*

### Why My Child?

Being told that your child has or may have tuberous sclerosis can cause a range of feelings. You may ask yourself, “Did I pass this disease on to my child?” or you may have fears for child’s future. These are common feelings as you learn to cope with this diagnosis. It is important to remember that everyone reaches acceptance at their own speed and in their own way. While one parent may quickly learn to accept the diagnosis, the other parent may have a prolonged feeling of disbelief and denial.

One of the most frustrating things about TS is that you never know what the next day may bring. Because TS is so variable, it is not possible to predict how your child will develop and which symptoms of TS he or she will have. You will have many questions—of yourself, your spouse, your family, your child’s physicians, and other parents who have children with TS. The uncertainty is sometimes difficult to deal with and puts stress on the family. Some parents have described it as feeling like walking through a minefield. Mutual support between both parents and any siblings is essential. Open and honest communication will provide strength for the whole family so that your child will have the support he or she needs. Participating in a TS support group (see the Support section) can be helpful for everyone involved.

Fear of the future can be immobilizing. But, as with any child, it is important to have high hopes and expectations! Your positive attitude, energies, and efforts will help your child to achieve all that he or she can. And, as with any child, many goals will be reached and many will not. Through it all, you will discover that your child enriches your life. The special love you give your child, the thrill of his or her progress, and the strength you will develop from dealing with this disorder may result in new discoveries for you.

The most important thing is to ensure that your child is receiving the best possible health care, education, and therapy, and lots of love from you. The appropriate testing and follow-up should help you understand what to expect rather than waiting to see what happens. One of the most important things you can do for your child is to learn as much about the disease as possible, be positive about your child's abilities and limitations, enjoy your child's accomplishments, and remember how much this child will bring to your family and to your life. Never give up on the hopes and expectations you have for your child, who is a very special part of your family.

### Where to Get Help

**Information.** Parents are encouraged to contact the headquarters of the Tuberous Sclerosis Alliance (TS Alliance) in Silver Spring, MD, for information; it can be found on the Web at [www.tsalliance.org](http://www.tsalliance.org). Or call the toll-free number at 1-800-225-6872. This line is answered Monday through Friday between the hours of 8:30 AM and 5:00 PM (Eastern Standard Time). An emergency contact is provided on the office voice mail when the office is closed. TS Alliance is a voluntary, nonprofit organization that is dedicated to finding a cure for tuberous sclerosis while improving the lives of those affected. To do this, TS Alliance fosters and supports tuberous sclerosis research; provides education of the public, educators, and health-care professionals; and provides support for individuals with TS and their families. The TS Alliance staff will answer your questions and send additional information as requested. TS Alliance also publishes a resource guide that contains additional information and lists resources available on a state-by-state basis.

Another source of information is the extensive network of volunteers called Area Representatives that exists across the United States, with at least one Area Representative in almost every



*Never give up on the hopes and expectations you have for your child, who is a very special part of your family*

state. Most of these volunteers are either parents of children with TS or have TS themselves, and they can provide information about local resources and can help you locate doctors in your area. They also coordinate local fund-raising projects. The TS Alliance staff can give you the name and telephone number of the Area Representative closest to you.

**Support.** The Area Representatives can also provide emotional support when you need it the most. These volunteers organize Parent Support Group meetings that can offer support and information for you and your family. Families living with TS can often help each other overcome feelings of grief, depression, fear, and isolation. It is important that you get the support you need to cope with the fact that your child has a genetic disease. At times, professional counseling may be advisable.

Another source of support is TS Alliance's quarterly newsletter, *Perspective*, which is available to all TS families. This newsletter will inform you about the current developments in TS research as well as new treatments and antiepileptic drugs, answers to questions about TS, tell you how other individuals and families cope with TS, and more.

In addition, TS Alliance has published a book entitled *Living with Tuberous Sclerosis—Stories of Love and Hope*. It contains 28 stories written by individuals with TS or their parents. This book helps paint a picture of the diversity of this disorder and of those coping with it. Through the contributions of generous donors, the first copy of this book is available free to TS families and health-care providers.

**Medical Help.** A pediatrician is usually the best source of general medical care for a child with TS. Referral to a pediatric neurologist may be necessary to establish the diagnosis and to

treat seizures or other neurologic problems. If your physician cannot make this referral, the pediatric or neurology department of a nearby medical school usually has a pediatric neurologist on its staff. It is a good idea to find a neurologist who has other TS patients and is familiar with the disorder. TS Alliance can provide you with a list of local families who may be able to help you find a neurologist or other specialist in your area. In addition, TS clinics across the country provide multidisciplinary care for individuals with TS and can arrange appointments with specialists. Physicians associated with TS clinics can offer medical advice about TS and its complications or can give a second opinion on the treatment and care of your child with TS. For more information on these clinics, please contact the TS Alliance.

**Special Education Needs.** Free public special education is mandated by federal law for any child from birth to age 21 experiencing learning or physical difficulties. Contact the Special Education Coordinator in your local public school system to have your child's educational needs evaluated at no charge. Refer to the TS Alliance Resource Guide for additional information about services and resources available in your state.

Because many children with TS have developmental delays, your child is in what is called a "high-risk" category. This does not mean that your child will have the most severe symptoms of TS. What it does mean is that the risk is greatly increased that some problems will occur in any of the developmental areas such as speech and motor skills. More than 20 years of research have shown conclusively that early intervention is very beneficial for handicapped children, their families, and society. We recommend that you get an early start on your child's education rather than taking a "wait and see" attitude. Even if your child's development does not appear to be delayed, intervention programs provide constant professional evaluation of develop-



ment and can catch any problems early, as well as give you peace of mind that your child is “on track.” Programs for infants and toddlers are called “early intervention” or “infant stimulation” programs. Through these programs you can learn how to help your child with his or her particular problems.

*Free public special education is available to any child experiencing learning or physical difficulties*

The professionals likely to be involved in evaluating your child for any special education program include an education specialist, a speech therapist, a physical therapist, an occupational therapist, and possibly a psychologist. During the evaluation, many “educated” eyes will be observing your child, but you should not feel intimidated. They are there to help you recognize possible problem areas and offer suggestions on what can be done about them. You will then be able to focus your time with your child where it will do the most good. The objective opinion of professionals will help you know what to expect of your child. Having a clear idea of what your child can do eliminates a great deal of fear. If you continue to encourage your child and keep a positive attitude, your child will achieve at his or her highest possible level. Remember that you will need to be your child’s advocate to ensure that educational needs are being met, just as you need to advocate for medical care. Be as involved as you can in forming the educational plan for your child, and ask as many questions as necessary to understand the process and what is being proposed for your child.

**To Learn More.** The following information may be helpful if you want to know more about TS. With continuing research and clinical developments in TS, some information will change significantly, so you should look for the most recent information.

- The European Chromosome 16 Tuberous Sclerosis Consortium. (1993). Identification and characterization of the tuberous sclerosis gene on chromosome 16. *Cell*, 75, 1305-1315.
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- Gomez M.R., Sampson J.R., & Whittemore V.H. (1999). *Tuberous Sclerosis Complex* (3<sup>rd</sup> ed.). New York: Oxford University Press.
- Hunt A. (1993). Development, behaviour and seizures in 300 cases of tuberous sclerosis. *Journal of Intellectual Disability Research*, 37, 41-51.
- Hunt A., & Stores G. (1993). Sleep disorder and epilepsy in children with tuberous sclerosis: A questionnaire-based study. *Developmental Medicine and Child Neurology*, 36, 108-115.
- Hyman M.H., & Whittemore V.H. (2000). National Institutes of Health consensus conference: Tuberous sclerosis complex. *Archives of Neurology*, 57(5), 662-665.



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- Nellist M., Brook-Carter P.T., Connor J.M., Kwiatkowski D.J., Johnson P., & Sampson, J.R. (1993). Identification of markers flanking the tuberous sclerosis locus on chromosome 9 (TSC1). *Journal of Medical Genetics*, 30, 224-227.
- Roach E.S., DiMario F.J., Kandt R.S., & Northrup H. (1998). Tuberous sclerosis consensus conference: Recommendations for diagnostic evaluation. *Journal of Child Neurology*, 14(6), 401-407.
- Roach E.S., Gomez M.R., & Northrup H. (1998). Tuberous sclerosis complex consensus conference: Revised clinical diagnostic criteria. *Journal of Child Neurology*, 13, 624-628.
- Sheffield C., & Sheffield P. (1999). *Living with tuberous sclerosis*. Landover, MD: National Tuberous Sclerosis Association.
- Van Sleghenhorst M., de Hoogt R., Hermans C., et al. (1997). Identification of the tuberous sclerosis gene TSC1 on chromosome 9q34. *Science*, 277, 805-808.

## Policy and Disclaimer

*This brochure attempts to describe information of interest to parents of children with tuberous sclerosis. Tuberous Sclerosis Alliance does not knowingly report inaccurate or libelous material. Tuberous Sclerosis Alliance does not promote or recommend any treatment, therapy, institution, or professional system. The information provided herein is not to be a guide for self-treatment. Follow your physician's instructions and consult with him or her on any questions, thoughts, or concerns you may have. Several organizations that may be able to provide additional information and assistance are listed below:*

### **Autism Society of America**

7910 Woodmont Avenue  
Suite 650  
Bethesda, MD 20814  
(301) 657-0881  
1-800-3-AUTISM  
[www.autism-society.org](http://www.autism-society.org)

### **Comprehensive Epilepsy Program and Epilepsy Information Service**

Bowman-Gray School of Medicine  
300 South Hawthorne Road  
Winston-Salem, NC 27103  
1-800-642-0500

### **Epilepsy Foundation**

4351 Garden City Drive  
Landover, MD 20785  
(301) 459-3700  
1-800-EFA-4050  
[www.epilepsyfoundation.org](http://www.epilepsyfoundation.org)

### **Learning Disabilities Association of America**

4156 Library Road  
Pittsburgh, PA 15234  
(412) 341-1515  
(412) 341-8077  
FAX: (412) 344-0224  
[www.lidaamerica.org](http://www.lidaamerica.org)

### **C.H.A.D.D.**

8181 Professional Place, Suite 201  
Landover, MD 20785  
(301) 306-7070  
800-233-4050  
[www.chadd.org](http://www.chadd.org)

### **National Family Caregivers Association**

10400 Connecticut Ave., Suite 500  
Kensington, MD 20895  
(301) 942-6430  
800-896-3650  
[www.nfcacares.org](http://www.nfcacares.org)



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(800) 225-6872

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