



## Talking to Your Child about Tuberous Sclerosis Complex

The diagnosis of tuberous sclerosis complex (TSC) may be made at any point in an individual's life, young or old. A newborn infant with white spots on her skin may be diagnosed with TSC when she has her first seizure and the tubers in the brain are seen on magnetic resonance imaging (MRI). A gifted and talented 5-year-old with facial angiofibromas may be diagnosed with TSC when his mother takes him to see a dermatologist because of the "rash" on his face. An adult may be diagnosed with TSC when his child is diagnosed with the disease. No matter when the diagnosis comes, it is a diagnosis that can be difficult to understand and accept, and it can be devastating for both the individual and the family.

### Why is this diagnosis so difficult for a family to accept?

First, it can be difficult to diagnose TSC, and it may have taken the individual with TSC several weeks or months, and visits to several physicians, before he or she received the correct diagnosis. In a survey of families associated with a TSC support group in North Carolina, the average delay from the onset of infantile spasms to proper diagnosis of the seizure type and of TSC was 65 days, and a median of three physicians were visited before the diagnosis of TSC was established (W.G. Ward, unpublished data, 1994). A neurologist made the diagnosis in 71% of the cases, and by a pediatrician in 21% of the cases. Only 57% of the children with infantile spasms were diagnosed by the initial physician as a seizure, and it was the most likely seizure type to lead to an inappropriate physician response to the family. Families may have been told that it was colic, indigestion, or gas, or just an over reactive infant. Inappropriate physician response to initial inquiries from the

family, usually the mother, can cause a breakdown in the patient-physician relationship that can lead to anger and distrust. Parker (1996) pointed out that, for many parents, the lack of professional support in their care-giving crisis began early. Most parents interviewed in her study stated that it was immediately apparent to them at birth that there was something different and possibly wrong with their child. However, physicians and nurses ignored their input.

Second, there is still a social stigma in some cultures toward individuals with genetic conditions. Some individuals will be told that they have inherited "bad" genes. Alternatively, people are just frightened by the unknown and do not know how to react to the diagnosis of TSC, or of a family member who has been diagnosed with this genetic condition. As we learn more and more about the human genome, we have come to realise that almost everyone has at least one or two mutations in their genes - some of these mutations will cause no problems, and others will result in either the diagnosis of a genetic condition, and/or will predispose the individual to developing a particular medical condition depending on their environment, the makeup of the rest of their genome, and/or other factors that we do not yet understand. As the diagnosis of genetic conditions becomes increasingly common, we will find a greater acceptance in the public of these diagnoses, as well as a greater understanding and compassion for the individuals with the conditions.

In the meantime, there are a couple of facts about TSC that are good to remember: 1) there is a tremendous variability in the clinical manifestations of TSC from one individual to another, even within the same family, when there is more than one individual with TSC. We do not yet understand why this occurs, but it is likely that it is due to the other genes each of us inherits that play a role in how TSC expresses itself. 2) The old literature about TSC, and unfortunately what is still taught to medical students, provides a view of TSC that is the worst case scenario. It is devastating to learn that your child has TSC, but when the

physician then cites literature that is out of date and proceeds to tell you that your child will definitely be severely affected, it leaves the parents in despair and with no hope. This is where it is important to educate yourself as a parent about TSC, obtain all the latest information from the TS organisation, and talk to other parents about their experiences. The advances in both the diagnosis and treatment of TSC over the last 10 years are astounding. There is every hope that your child will have a wonderful and productive life.

## **How do I become educated about TSC?**

The best way to become an informed and educated parent about TSC is to obtain information from the TS organisation, join a support group if there is one close to you, and/or talk to other individuals with TSC and/or parents of children with TSC to learn from their experiences. It is often the case that the parents will know more about TSC than the child's physicians, especially primary care physicians. The parent should remember that this might be the first child with TSC that the physician has cared for. It is your role as a parent to provide information for the physician and to find a physician that you can collaborate with to provide optimal health care for your child. Your child will be provided the best care possible if you find a physician who is willing to learn with you, who is willing to talk to other physicians, and who is willing to tell you that they don't know the answer to your question, but they will help you to find the answer.

TSC is a complicated genetic condition because it can involve so many different organ systems. It is important as a parent to focus on the issues at hand while anticipating the issues that may or may not occur in the future. Current recommendations for the initial evaluation at the time of diagnosis and for follow-up screening should be followed so that issues that may develop down the line are followed and treated as appropriate rather than taking a 'wait and see attitude' that may result in a health crisis.

## **Should I tell my child he/she has tuberous sclerosis?**

The answer to this question is yes, but the right time to tell your child will depend on the family dynamics, the child's abilities and maturity level, and the clinical manifestations of TSC seen in the

child. In an informal survey of 10 families with children with TSC, each family had their own unique approach to telling their child that they had TSC. Most of the families indicated that the best time to tell the child was when they were very young so that they grew up knowing that they had TSC. Some parents regretted not telling their child when they were younger because they felt they could have spared their child from being fearful of the doctor's appointments and unknowns about their condition if they had told them earlier. Several individuals indicated that telling a teen they have TSC was not the thing to do! The teen years can be especially difficult to start with, but then having to tell a teenager that they have a genetic condition can be devastating to them at a time when their self-esteem may be suffering already.

Several parents indicated that answering the child's questions in as simple and straightforward a manner as possible was the best approach. The child may be happy with just knowing that there is a name for their condition, and that this is why they have to go to the doctor. Explaining that it is important to get a "picture" of their brain, and the only way this can be done is with either a MRI or a CT scan, may help them to understand why these tests are being done.

One parent indicated that it helped to talk to her child about other individuals with TSC so that the child began to understand that there are many forms the condition could take. The child would ask, "What kind of TS does he have?" when the mother referred to an individual she knew. It provided the opportunity for the mother to tell the child what this individual's clinical manifestations of TSC were, and to then tell the child that they do or do not have these same manifestations. Introducing a child with TSC to other individuals with the condition can also be extremely helpful. It helps a child feel that they are not alone with their condition if they know that there are other individuals out there who are living and coping with TSC.

## **Resources**

Many web sites have been established for children with genetic conditions. One good site with links to others is [www.tarynsworld.org/](http://www.tarynsworld.org/) (Taryn has Mannosidosis). A good web site by an adult with TSC is Lisa's Site: [www.title14.com/ts/](http://www.title14.com/ts/).

## **Should I tell the other children their sibling has TSC?**

Research has shown that it is important to inform the siblings about the diagnosis of their brother or sister, and to make sure that their needs are being met. It can be stressful and difficult for the sibling to comprehend what is happening to their sibling if the child has frequent hospitalisations and/or visits to the doctors. A young child may be frightened that they caused their sibling to get sick, or that they themselves will get sick and have to go to the hospital. There may also feel neglected if the child with TSC has chronic health needs and requires a lot of time and attention from the parents.

As with the child with TSC, it is important that as parents you are honest and straightforward with the siblings. They need to understand that they cannot “catch” tuberous sclerosis from their sibling, and that they were not responsible for their sibling’s condition. If they are brought into the process, understand that their sibling has special needs, and they are a very important part of their sibling’s life, they will accept from a very early age the challenges placed on them and their family. This is not to say that it will always be a smooth road - there will be times when the siblings feel neglected, angry, or concerned. It is during these times when it will be important for the parent to give some special attention and time to the siblings so their concerns and needs can be attended to.

There are excellent resources available on the Internet for siblings. Donald Meyer has collected these resources on the following web site: [www.seattlechildrens.org/sibsupp](http://www.seattlechildrens.org/sibsupp)

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**Tuberous Sclerosis Association**



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