

In Their Own Words

*Personal Journeys
with Tuberous Sclerosis Complex*



Tuberous Sclerosis Alliance

Foreword

“In Their Own Words...Personal Journeys with Tuberous Sclerosis Complex” was compiled to give readers a glimpse into the lives of ordinary people and families who face the challenges of tuberous sclerosis complex (TSC) on a daily basis, whether they are infants, children, teens or adults. Because TSC symptoms range from mild to quite severe, stories were chosen to reflect a wide variety of experiences. These heartfelt journeys on the road of life offer inspiration and hope not only for their own families, but also for anyone affected by TSC. We hope you find solace in knowing you are certainly not alone.

Please note that many of the stories discuss specific treatments and/or use of certain medications. The TS Alliance does not endorse or recommend any specific treatments, health care professionals or hospitals or institutions such as those mentioned in the booklet. Please refer any medical questions directly to your health care professionals who are in the best position to offer medical advice regarding your health care.

The TS Alliance is grateful to all the authors, families and individuals who contributed their personal reflections on living with TSC.

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Signs of Life: Another Perspective on Tuberous Sclerosis Complex and Autism

by Donna Satterlee Ross

Down the street from my mother's Florida home sets a billboard with a picture of two vibrant looking seniors that proclaims to the world in giant-sized letters, "This is living!" The sign advertises a community boasting every type of luxury and entertainment imaginable, promising anyone who resides there a fun-filled life of comfort and ease...a dream come true. And yet each time I pass it, I find myself thinking, "*Is this living?*"

While we are constantly bombarded with images like these, and we are surrounded by reminders that the "good life" means "having it all"—the biggest house, the perfect family, the fastest car, and the flattest TV, the actual definition of living is quite different. The Oxford Dictionary of Current English states that life is: the capacity for growth, functional activity and continual change until death. Real life, it seems, does not come with 2.5 children or a maintenance-free exterior.

So what does it mean to truly live? Well, last year on Easter Day my infant daughter, Adria, was diagnosed with tuberous sclerosis complex (TSC), a serious genetic disorder that would affect her development throughout her life. Instead of enjoying dinner with the family, we spent the weekend at the hospital learning about the tubers in her brain, which would likely cause her to face on ongoing battle with epilepsy, intellectual disability and autism. Suddenly our family was left wondering how her life would be—how would we provide for her needs, which were great? My husband and I honestly didn't know if it was better to hope for her to live or die.

Then one day shortly after the diagnosis, I was walking with Adria on a trail near our house and instead of passing by the cemetery on our route, I decided to detour through it. As I made my way down the pathway, I noticed a statue of what appeared to be a small child and a dog. The writing on the sign was very small so I had to get even closer to read the words on the sign, "BABYLAND." I'm sure my gasp would have been audible if anyone else had been nearby. Carefully, I stepped closer to examine a tiny grave marker, placed so near to the next. I was overcome by indescribable emotion as I read the names and dates. The first child had lived a day. The next lived only a few months. Then came a much larger marker—quadruplets. I could not even imagine the grief that family must have felt. And yet the messages were simple and sweet: "Too good for this world." "God's Littlest Angel" "We'll see you again soon." The markers proclaimed the sentiments quietly, but with a power that could never be equaled, even by the largest sign.

I found myself crying—not tears of sorrow, but tears of hope. I experienced a renewed sense of gratitude as I realized the bundle I was carrying in a pack on my back was not just a precious child, but indeed life itself. With the capacity for growth and change (however slow), with a smile in the morning and kisses every night. Like each of us, striving to make an impact on the world and those around us (however slight). Today I can make my own claim, "*This is living!*"

May we each approach the year with renewed life!

Christian Joel Martinez

By Lucy Martinez

What started out as an ordinary day turned into one we will never forget. My pregnancy was pretty much uneventful until the last trimester when I started having swelling from fluid retention and my blood pressure seemed to be increasing. I was two weeks away from my due date and went to a routine OB checkup. Because of the increased swelling and rising blood pressure, the doctor thought it would be a good idea to put me in the hospital for observation, an ultrasound, and what I thought would be a possible cesarean section due to toxemia.

As I lay on the table with an ultrasound tech holding the wand in one spot with a funny look on her face, I knew something was wrong. I don't remember saying anything, but when she left the room to get a radiologist, I began to worry. I told the radiologist that since I'm a nurse, he could tell me what's wrong. He said something along the lines of, "I see something in the baby's heart, but it could just be static; it may be nothing at all. I will call your doctor."

The next thing I remember is sitting in a wheelchair in the hallway, teary-eyed, not knowing what to think. I went to another facility for a high-level ultrasound. "Boom," said the doctor, "he's going to be a big baby and, oh, he might have tuberous sclerosis complex (TSC) and rhabdomyomas." I remember balling on the table, because he started talking about open heart surgery, and I didn't have a clue about what TSC was. I had him write it down on yellow sticky. I went home and waited.

As I frantically tried to look up these foreign words on the Internet, my OB called to tell me to come back to the hospital so I could be transferred to a larger facility with a higher-level nursery care. My husband and I went, not knowing what to expect.

Three days later on June 1, 2003, at 11:07 p.m., Christian Joel Martinez was born by C-section. After a whole team of staff looked him over—and a quick kiss from me—he was whisked off to NICU. I didn't get to see him for another 24 hours, when my medication had worn off and I was able to move a little. I finally saw my precious baby boy, who looked perfect but had to be put through a battery of pokes and tests. The whole week went by with him seeing specialist after specialist. Probably the most important one was our geneticist, Dr. Hope Northrup, who confirmed the TSC diagnosis. We were also now armed with a lot of information but didn't know what to do with it.

After a week, Christian and I went home. We watched and waited. At eight weeks of age, little jerky movements started; the expected seizures had begun and a seemingly never-ending journey to multiple doctors had also started—I never saw so many doctors in my life. I know he had to feel traumatized by all the testing, sticking and hospitalizations.

At 13 months of age, Christian had brain resection surgery to remove a large tuber in the right frontal lobe where all the seizure activity originated. We didn't stay in the hospital long, which was good because although Christian breezed through it all, we were having nervous breakdowns.

We never witnessed further seizure activity, and it was time to get him back on track developmentally. We started physical, occupational and speech therapies. At 17 months of age, Christian started sitting and later crawling came, which was rough on his knees because he would crawl a long time before we saw him trying to pull himself up. Words like “mama” and “dada” also started coming. I remember being more excited than ever when he picked up a “Cheetos” to put in his mouth, which was one of those little things most people take for granted.

One day during physical therapy, the therapist told me to come see something. I couldn't believe my eyes: Christian was walking using a tiny walker! It took awhile, but at age two and half he started walking! I know some of you can share my excitement.

As Christian worked on his strength and balance, we needed to get more aggressive with his speech. We started using sign language, and he started to catch on so we were able to communicate a little better. Otherwise, we went on with our busy lives of working and taking Christian to the sitter, doctor appointments and therapy sessions. He continued to learn more signs and say more words.

When he turned three, Early Childhood Intervention helped him make the transition to start school in August 2006. His speech took off so much that we stopped using sign language. He says everything (or rather his own version) and is now able to put three words together. Of course, he isn't caught up to his age level, but we are pleased with his progress.

He gets private therapy as well as therapy at school. In December 2006, we took him to a psychiatrist because of high anxiety levels, and a new medication was prescribed. He is doing better now, but at the time, he had every reason to be scared when we went somewhere strange—he thought we were taking him to the doctor or hospital. Sometimes, he just doesn't want to be in a particular place so he acts out. But this seems to come with the territory, and we are working on it like everything else.

Christian is loving, sociable, funny and very cute with his glasses. He says “please,” “thank you,” and “bless you” when someone sneezes! He just picks up things on his own, and sometimes he amazes us with his memory. We can usually tell him something one time, and he will remember it because he's so inquisitive.

We would not trade anything we have had to endure over the past few trying years for something else. It has made us a stronger family and has opened our eyes to a different world. No one will ever know what the lives of TSC families are like without going through the experience themselves. I am more compassionate toward other disabled people I encounter, and I have met the most wonderful families locally who are also dealing with TSC. We are able to work for our cause through walkathons, other fundraising events or just being there for each other.

Since I started writing my story, things have changed in our lives though. Christian started having short staring spells to being “dazed out” during longer staring spells, his lips turning purple, then getting sleepy. We haven't witnessed these occurrences every day and there have only been a few, but we had to increase his medication again.

My faith plays a tremendous part in my sanity. Don't get me wrong—I do have an occasional meltdown when I'm alone, but I can do anything as my spirituality strengthens me. I embedded that in my brain awhile back. I believe Christian was a gift to us and that we were chosen. Christian gives us unconditional love and is innocent in all of this. We, his parents, will do everything in our power to make sure he is given every opportunity to grow, learn and become as independent as possible. I pray that newer parents will have it even better through new technology, improved medicines and procedures, and one day a cure.

My Dear Sweet Anaston

By Elizabeth Burnett

My name is Elizabeth. My husband, Richard, and I have three beautiful, energetic children. Noah is now six and loves sports, hunting and anything outdoors. Abram and Anaston, our twins, are now almost three years old and love playing with each other and the outdoors as well. We feel truly blessed and thankful every day for our children. However, three years ago our situation was a bit different.



Noah, who was two-and-a-half years old at the time, was our only child together. When I married Richard, he already had two older boys, but we decided to try to have one more child closer to Noah's age. When the doctor told us we would have twins, it was a shock but nothing like the shock I received at my 27-week ultrasound. "You know," the technician said with nervous smile, "it's probably just shadows I see on the little girl's heart but I want to make sure, so I will send you to have another ultrasound in the morning with a high-risk doctor present."

I came home that day and cried to Richard that something was wrong with my baby girl's heart. I realize now if the tumor had not been detected, Anaston would have simply died in her sleep because of the lack of blood flow to her heart, the death would have just been labeled SIDS. However, that was not part of the plan. The next day we heard it for the first time: tuberous sclerosis complex (TSC). We asked, "What's that?"

So we came home to look it up on the Internet, which was probably a big mistake, because we found mostly outdated information that made us panic. We didn't know about the TS Alliance at this time.

My twins were born full term at Duke Hospital. Because of the blockage from a tumor on Anaston's heart, open heart surgery was successfully performed three weeks later. At three months of age, simple partial seizures began. Anaston walked on time (at 10 months) but her fine motor skills and speech were delayed.

The first year was the hardest. Doctor appointments and not knowing what to expect left us feeling very anxious at times. I am so thankful for the support of family, church members, friends and most of all my spiritual guidance, which I believe lead me to write a poem during that time. My pastor let me read it on Mother's Day 2006. I write it here as well, sharing from a Mother's heart:

Dear Sweet Anaston

*Will you ever know, The Blessing you are,
From blonde hair to sweet toes.*

*I will never forget the day we found about you.
We cried and felt so lonely and blue.*

*Little did I know God's plans for me,
My husband, our children, family and friends.*

*Anxious as I was about your arrival,
You see they told me there wasn't much hope,
for your survival.*

*They, again and again, told me just how sickly
you would be,
And how things would be better just to end
the pregnancy.*

*That's when your daddy and I realized,
God was the only one, to handle problems
this size.*

*I remember bowing on my knees to pray,
"Why our little girl? Why us? Show us the way."*

*Jesus where are You? Let me feel your presence,
I would pray,
Then that's when Noah said,
"Mom, I see Jesus, He's in this house today."*

*Thank you God for sharing me that day,
I love you dear Father is all I have to say.*

*Now go with us to Duke and be with us through
this delivery.*

*Born into this world without a heart beat,
Not a sign or whimper, but wait a minute,
you were pink!
Her cord is wrapped too tight around her neck,
I heard them say, Get some scissors and
don't delay.*

*How could this be, the doctors all thought,
A child this sure of death...Wait a minute, there's
a cough.*

*Then a scream and a loud one too,
Wait, that's not just Abram,
That's his sister and why is she not blue?*

*Sometimes we go through our whole lives not realizing where our blessings come from. God
worked one of ours through a 5 lb., 7 oz. Angel named Anaston.*

*Thank you God,
Elizabeth and Richard Burnette*

*As pink as you could possibly be,
Mouth open wide screaming at me.*

*I knew right then God was in charge, and
had been all along.
He has proven each and every one of them wrong.*

*Oh, what beautiful babies we have, I'm so happy
it's over,
So exhausted, but so glad.*

*So many things happened since then, you see,
Your seizures, small delays, and your open heart
surgery!*

*But, through it all I have to say,
God has helped us through each and every day.
You see sometimes a child comes along that's
labeled,
Even before birth, as special or disabled.*

*The only thing that I can say, is that each
one of my
Children are special in their own way.*

*Given to Richard and myself, here on earth to
love,
Knowing they're really our God's up above.*

*He thought of Richard and me, enough to know,
That with His help, our children will grow.*

*Into adults that were raised, by Christian parents,
That will always give praise.*

*For our dear Father above,
His sweet miracles, healing and love.*

*So sweet Anaston you must know,
that God will be with you, each and every day
you grow.*

*Into a beautiful woman, that I know,
Will continue to bless everyone, everywhere you go.*

*Thank you God for using our daughter,
Working through her to save me and her father.*

*For without her, we would have never been,
So close to You and still living in sin.*

This poem has now been passed on to other people with special needs children or just problems of their own. The poem has been a blessing to many but it wouldn't have been written without my faith guiding me, my daughter Anaston or yes, TSC. No, I do not like the fact that Anaston has TSC, but she is Anaston first—TSC is not my daughter; she just happens to have the disorder. Before her, I believe we were self-centered, self-reliant, less compassionate and far from having any type of faith. Today we are selfless, totally dependent on our spirituality, and very compassionate, which makes us and our children as better all-around people.

Very recently Anaston had the vagus nerve stimulator implanted, which I believe is another miracle resulting from my faith. Today, she is thriving and smiles and plays all the time. She loves her family and of course playing with all of her brothers. She is a true angel.

We have no idea what the future brings for Anaston. We know things can develop from this disease that could affect different organs. But, we have no idea what the future brings for any of us. We just take it day by day and love and enjoy our children and the different challenges and joys that each one brings.

Gabriel's Story

by Elizabeth Hallauer

This is Gabriel's story, and I thought you might be interested. My son, Gabriel, was a perfectly healthy child until he was six months old. He began to have some seizures...his little body would tense for several seconds. We rushed him to the local hospital and the ER did a blood test. The doctor came in to tell us we could go home since she had not seen Gabriel's seizure.

I took him to see his family doctor the next day, and he decided to have an EEG (a brain-wave scan) done. As we were waiting, my son had another really bad seizure. We went to Vanderbilt Children's Hospital immediately. They checked him over and ran some tests, but couldn't schedule an EEG until Monday so we decided to go home. This was the longest weekend ever for all of us. Monday the EEG was abnormal, so they put Gabriel on seizure medication. At this time they thought it was epilepsy. I still felt like there was something else going on. The next day he had a horrible seizure that put us back at Vanderbilt. This time I asked that he be observed overnight. The doctor agreed and ordered an EKG done as a safety precaution. We spent the next few hours sleeping when we could in straight, hard hospital chairs. Gabriel was finally admitted for observation. The next morning it seemed like doctor after doctor came and did test after test.

They finally diagnosed him with tuberous sclerosis complex (TSC). This is a pretty rare disease, and one in every 6,000 live births will have this genetic mutation. My son has benign tumors in the brain, heart, kidneys, and eyes. He is taking seizure medication and having therapy two days a week. It is truly one day at a time with Gabriel. However, he has a smile that will melt your heart and a spirit that will light up your life. He is our little blessing!



Kylie's Hope

By Maria Gibbons

My entire life changed the day I became a mom. I was handed this beautiful, perfect creature, who had deep blue eyes and swirls of blond curls. As she curled her little fist around my finger, I was hooked. I gave her my whole heart and entire soul, in one tiny instant. I dreamed of the two of us playing dress up, going shopping, having tea parties, and putting make-up on each other. I imagined showing her how to throw a fast ball, how to drive a car and watching her walk down the aisle, as she married the man of her dreams. I also pictured her holding her daughter in her arms and thought, life couldn't get more perfect than this.



Kylie was 4 months old when she had her first seizure. We were lying in bed together one morning, when she started sticking her tongue out. She was moving it in and out, in a rhythmic motion. Her eyes started deviating to the left and then she fell asleep. I stared at her, in shock. Being a nurse, I knew she just had a seizure, but my heart wasn't ready to accept it. I took her to the doctor, where she checked out just fine but they sent us to a neurologist, more so to calm my "new mother jitters." She checked out fine there as well, and we were sent on our way. Life was good again.

Two weeks later, Kylie was rushed by ambulance to Children's ER in St. Paul after experiencing another seizure, this one lasting longer and scaring the heck out of all of us. She received Phenobarbital, making my baby literally unconscious for two days. We were brought up to the pediatric epilepsy floor and I remember thinking, we don't belong here. There were kids in wheelchairs, wearing helmets and drooling. My baby just had a reaction to her baby shots, and we would be going home soon. How naive I was and how I wish it could've been that simple.

I remember the pediatrician and neurologist coming in to talk to us, stating ever so kindly that they had a suspicion Kylie had tuberous sclerosis complex (TSC). An MRI showed two tiny spots on her brain, but the scans were being sent to a few different neurologists across the country. One day, a team of doctors came into our room. From that moment on, I knew our lives would never be the same. My heart broke into a million tiny pieces as I cried for my baby and grieved for the life we would never live.

It has been just over two years since Kylie was diagnosed. Some days it feels like we are on a roller coaster. We have had some really, really, really good days and an equal amount of really crappy ones. Kylie had a two-month break from seizures when she was 14 months old but they came back with a vengeance. We spent a month in the hospital, where the seizures finally stopped cold turkey. She went from 17 to 27 months of age without a seizure, and I had days when I almost forgot she had TSC.

She finally started to walk at 26 months, and I can't even begin to describe the joy in my heart. She worked so hard for so long and she did it! There is nothing more magical than seeing your child achieve milestones for the first time, and I think because we had to wait for so long, it was even all the more special. She was doing so well that she was weaned off a seizure med (she was on five medications at the time). Unfortunately, we chose the wrong one to wean. She ended up back in the hospital, seizing nearly every three minutes at one point in time and having roughly 100 seizures a day. After two weeks, we went home and the seizures have once again stopped. We pray every day they will continue to stay away but realistically, we know they can come back at any moment.

TSC has changed my life in so many ways. I have come to the realization that life is a gift—it can be altered or taken away at any given time. We do not get to choose the life we are given but we can make the most of what we have.

Am I angry that TSC has robbed my family of normalcy? Of course. I long for the ease of the lives my friends lead, one that doesn't involve having a child sleeping in between my husband and I, one where I wouldn't have to turn around and drive home because I forgot the Diastat®, one that didn't involve weekly doctor appointments, daily therapy appointments and the mass chaos of trying to keep Kylie's paperwork all organized. I would be totally ok without knowing how to decipher what an EEG recording says, to not know what all of the different seizure types are and the names of all of the top TSC specialists names in the country.

I can name all of the seizure meds out there, even the ones in clinical trials and what the normal lab values should be for a child. We should be going to dance class, not therapy appointments. We should be running in the grass, not falling on our hands and knees. We should be shushing Kylie for talking too loud, not clapping for each new word she says.

But do you know what we are doing? We love our sweet girl, each and every day we have with her. Kylie has a smile that could light up the darkest room and an infectious giggle that can only make you laugh with her. She is kind, gentle and beautiful. We may be falling every few feet we run, but when we are sitting in the grass, we are exploring ant hills and lady bugs. We are splashing in puddles just because it rained and cheering her on whenever she says something new. Every day is a party at our house.

We have met some of the most courageous and inspirational people that I now have the privilege of calling friends. We have learned which friends will be on our doorstep at 1:00 in the morning just to wipe away our tears and which ones just won't. I have seen so many different people come together to pray for us; some I would have never met if Kylie would not have been sick. I have learned that I am much stronger than I would've once thought and that the love of family is stronger than anything else in this world.

I don't know where we will be 10 years from now, five years from now, or one year from now. I don't even know what tomorrow will bring. I do know that I have an amazing, strong and beautiful daughter, despite TSC. It does not define who she is or who our family is.

Every time Kylie wraps her arms around me and whispers, "Ma-ma," I melt. When she blows kisses to me as I am leaving for work, I know I will have a good day. She has brought many blessings into our life and I wouldn't have it any other way. We continue to have hope for her future and that tomorrow will continue to bring smiles, giggles and sunshine.

Matthew Carlos

By JoAnn Carlos

Matthew will soon be 12 years old, and these past years since the TSC diagnosis have been a challenge, yet getting to this point is a relief. He has really grown and changed and now that we have found the help needed with children like this, we are doing great.

Matthew is enrolled in a very good school that concentrates on interaction with other children. Since Matthew was diagnosed with autism as well as TSC, they are working on the skills he will need later in life.

His seizures have been well under control with medication. He is currently taking Zonegran®, Tegretol®, and Depakote® for seizures; for behavior, he takes clonidine. He has about 10 complex partial seizures a month, which is great considering he used to have about 10 a day. He has grown to be a big kid; he's 5'3" and about 130 pounds. He loves to swim and is currently enrolled with the Special Olympics. However, I think Matthew just likes to be in the water. He doesn't seem to care what's going on with him when he's in the water. He has always been like that, even when he was a baby.

Matthew hasn't had any surgeries except having his gallbladder removed, which didn't seem to be related to the disorder. Even after that surgery he bounced back after two days. Pain really isn't a factor with him, which is really scary, but he seems to express that something is wrong through other moods. He tolerates pain and is very strong. When he doesn't get what he wants, he really can be stronger than a grown man.

Matthew still loves to be chased like any other normal kid, but he found a new love with stickers. They seem to occupy his hands and help with the coordinating and picking up small objects. He still doesn't talk, but when he is told to put shoes on, or something like that, he knows exactly what to do. Having a routine is a major part in his learning process. Without it, Matthew seems lost.

Matthew still has no verbal skills and still wears diapers, which we are working on a lot. I think toilet training will be the hardest thing to teach him because he still doesn't get the concept. He does feed himself but only certain foods. Enchiladas are his absolute favorite meal, and he can eat that every day, morning, noon and night!

Now that Matthew has been really good at interacting with other children, we find that it is time for him to enjoy being a big brother. He will be a big brother in June to a baby girl! He has shown that he can love and be affectionate so I can't wait to see what this new chapter in his life will bring us all. Matthew has lots of friends and help since he is really loveable.



TSC Is Only A Part of Our Lives

By Beth Tinlin

Our story involves three family members with tuberous sclerosis complex (TSC). We have had our years of major TSC issues, but all in all, today I can look back and say, "TSC is a part of our life, but it is NOT our entire life." We learned this due to the TS Alliance, its online community family, and the wonderful care we receive.

I can still remember the day the doctor told me straight out, "The baby you are carrying has six rhabdomyomas in his heart." In that moment, our world changed forever. I went home crying to my husband. I was seven months pregnant. The same week, our eight-month-old son Nathaniel was also diagnosed with infantile spasms and TSC.



That was the beginning of our TSC journey. Nathaniel had been having infantile spasms since day one of his life; we just did not realize what they were. I gave birth to Shane, our fourth son, that December. The doctor came in two hours after I delivered and said, "He has more tumors in his heart than we can count so we will monitor him in NICU for two days and then release him. If he lives a week, bring him in for a checkup."

We took Shane home two days later with an apnea monitor that went off more than 200 times the first week. It sounded like a fire alarm and you couldn't shut it off. Every time it went off, Nathaniel had a seizure, and our older sons, Zach and Nevada, came running thinking their brother was dying. We then went for the one-week checkup, and the alarm sounded in the doctor's office. We still could not shut it off; needless to say, the doctor switched monitors after that.

Nathaniel and Shane went through all the TSC tests about the same time. There I was with two babies undergoing brain MRIs, echocardiograms, EEGs, kidney ultrasounds, etc. Nathaniel was diagnosed with cortical tubers of the brain, infantile spasms, rhabdomyomas in this heart, and severe developmental delay. Shane was diagnosed with too many rhabdomyomas in his heart to count and lived the first six months of his life on a hospital heart monitor. Shane did not have brain, kidney, or lung involvement.

My husband, who has TSC, went through the guilt phase of "what did I do?" Looking at Doug, you would never know he has TSC, other than some skin markings. He does have one cortical tuber on the left side of his brain and the skin markings on his face and legs. He also has attention deficit disorder.

Shane's heart rhabdomyomas disappeared after almost two years. He never skipped a beat or had any blockage from them. Shane's only sign of TSC was not speaking. We started early intervention with both of our boys when they were babies, including learning sign language through the American Education Association.

At age three, Shane started speaking one day and has not stopped talking since. We call him our “information booth”—if anyone wants to know anything, we ask Shane. From the time someone walks in the door he tells us who did what, who called, what was said, etc. During Shane’s yearly MRI of the brain, some subependymal nodules showed up a couple of years ago. Currently, Shane has ADHD, ADD, and a new heart rhabdomyoma that two doctors swear were not there before. He still does very well, attends second grade in a regular classroom although he uses special resources for a delay in reading, comprehension and auditory processing issues. However, Shane looks like a normal but hyperactive eight year old.

Nathaniel was hit the hardest in our family early on with TSC. I am very grateful for the TS Alliance, particularly its online discussion group. I learned so much from the online veterans, who helped me realize the bitterness I felt needed to be refocused into advocating for my sons and for showing so much patience and guidance on what to do.

For example, I learned how to get vigabatrin (Sabril®) for my son. Nathaniel had infantile spasms up to 100 times every day for three and a half years. A mental evaluation at a little over three years showed him to be at a 10-month level. We started vigabatrin, and in 24 hours he went from too many spasms to count to zero. He then started to walk, talk, eat real food, learn his ABCs, numbers, etc. Wow, what a difference! I now understand why some neurologists say it is the number one recommended drug for infantile spasms in TSC children. It was our miracle drug.

This honeymoon period lasted about one year. We then switched to a neurologist in our city that we had heard so many good things about. Some of the most important things I learned from the TS Alliance’s online veterans are to listen to your gut; write everything down (i.e., number and time of seizures, actions taken, etc.); and remember that you, the parent, are more familiar with the child than anyone. I kept telling this doctor that we were seeing more and more seizures of different kinds, and the medications were making Nathaniel worse. Nathaniel became psychotic on top of the seizures, while being on seven antiepileptic drugs (AEDs) at one time. He would yell, scream, curse, and his favorite saying was “I hate you mom,” while biting, pinching and hitting us.

At that point, we decided enough was enough and started weaning him off all of the drugs, except vigabatrin. We also went back to the neurologist at Children’s Hospital of Iowa, which was the best decision we could have ever made. It took about 14 months of trying different AEDs to identify one that could control Nathan’s severe complex partial seizures. Finally, Trileptal® worked. After that, the psychosis and behaviors went away, and we had our handsome blonde-haired, blue-eyed smiling son back. Nathaniel has failed 12 AEDs, and brain surgery or vagal nerve stimulation are not options for him due to his heart condition. So, it is one day at a time, and we thank God for the honeymoon periods that last months now in between seizure times.

Nathaniel was about four when his heart started skipping beats. He was diagnosed with tachycardia, which turned into bradycardia and second-degree heart blockage. In 2004, we rushed him to the hospital for emergency heart surgery. They did a cardiac ablation. I can remember waiting for the surgeon, Dr. Law, to fly in from South Dakota, thinking this can’t be happening. Doug and I sat in that hospital all alone, missing Zach’s first football game of the year, and just asking why? We were waiting at the door to the operating room when the doctor came out to tell us it would all be okay. That surgery did not fix Nathaniel’s

heart problem though; it took a second surgery— a thermal catheter ablation performed by Dr. Law in 2006—to correct the problem. Following that surgery, Dr. Law was thrilled to tell us it was successful and to remove Nathaniel from all heart medication.

Nathaniel is currently nine years old and enrolled in a special education class on a limited schedule. Our nurse accompanies him to school. He reads, writes, and does math at about an early second grade level. His seizures are mostly controlled, with a few breakthroughs here and there, but his heartbeat is beautiful to listen to and his Tourette's and other behaviors are mainly controlled by medication.

Shane attends regular classroom at grade level with a health aide. He has migraines, ADD and ADHD, but otherwise does very well. Every six months, we continue to monitor his subependymal nodules. He also had a couple of absence seizures recently that he never experienced before.

It has been a long journey that is not over. The things we try to remember are to take one day at a time and don't worry about tomorrow. We also don't let TSC rule our entire lives because it is only a part of it, not all of it.

No, it is not easy to live a life filled with things like individual education plans at school and outcome achievement plans for respite, nursing and support services. We have needed medical and other kinds of support to help in our home for about the last seven years, making us feel like we live in a fishbowl where everyone can see what happens in our home.

My advice is to find medical care and other support you are comfortable with by listening to your instincts. Our primary care physician makes my life so much less stressful by writing letters of support for our individual education plans, obtaining services for our sons, listening to the boys about what they feel, and truly caring about what happens to them.

Also, contact the TS Alliance for information and support. Consider using the online discussion groups. Advocate for your children by fighting for services that will help your children to be the best they can be. Most of all, remember some family members may not really "get it" but they love you and can support you. We could not have survived this without the support our family or the online discussion group veterans.

My last piece of advice is to find care at a TSC clinic if you can. If you are unable to do so, don't beat yourself up over it. Just find the best care possible. We really feel our doctors are the best of the best—they listen, they learn with us, and they care. That is what medical care should be.

I can honestly say I feel we are success stories of surviving a life with TSC. We have been so blessed to have our family, the TS Alliance, doctors who truly care about our children by giving them the best quality of life, and most of all the veterans of the TS Alliance's online discussion groups who guided me to being a parent who advocates in every way possible for my sons and husband.

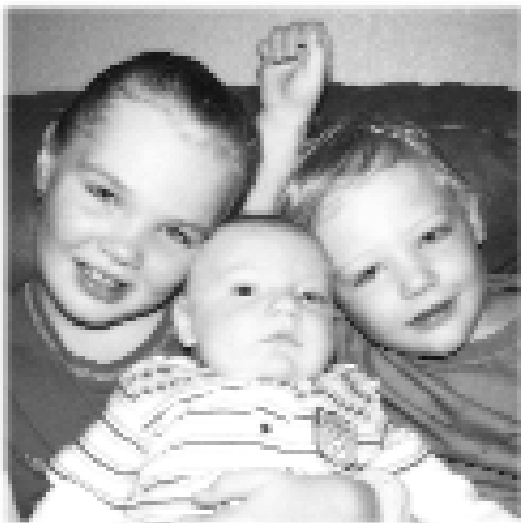
My “Picture Perfect” Life

By Shannon Hanks-Grandia

Growing up, I had lots of dreams about how my life would turn out. I would find a career I loved, get married and have a couple of healthy, happy children. We were going to be the picture perfect family, white picket fence and all. I was prepared that there might be some trials along the way but overall, life would be problem-free. If you had told me then the path planned for my life, I would have told you that you were sadly mistaken.

I did find a career that I loved in teaching, and I did marry the man of my dreams—my high school sweetheart, Rob. Unfortunately, that is where my picture-perfect dream is altered. Rob ended up having a

genetic disorder known as tuberous sclerosis complex (TSC), which was not diagnosed until our first two children began having seizures and were then diagnosed. Most would think that the trials would end there, but then my third child came along and he too has the disorder. By most people’s standards, we are far from perfect, but I would not trade my life for anything. I love the picture of my life and the path my faith continues to send me on.



Our first child, Rylee, was born in March 2001. Rob and I were ecstatic as she was a healthy and happy child. At the time, I was finishing training to become a teacher, and Rob began working for a company he loved. Rylee was cared for by my mom, her beloved Grammy, during the day. My picture-perfect life seemed to be falling into place.

When Rylee was 18 months old, I took her in for a checkup and asked the doctor about some white spots on her skin and some staring spells she was having. Rylee’s doctor didn’t seem concerned and referred us to a dermatologist to look at the spots, who said the white spots were one of two things, Vitiligo or another disorder she didn’t want to tell me the name of because it was “too scary.”

After a trip to the ophthalmologist to help rule out the “scary” disorder, Rylee’s staring spells began to worsen. Her eyes were now rolling back, and she was salivating during the episodes. We called Loma Linda Pediatric Hospital and were told to bring her in immediately for seizure treatment. After numerous tests, we were told that my baby girl had that really “scary” disorder and it was called tuberous sclerosis complex (TSC). This is when I felt like my life was being turned upside down.

I was a young mom who was shocked by the diagnosis. What did this mean? What was

going to happen to my baby? I remember crying in the courtyard of the hospital, scared and confused. I didn't feel angry, but overwhelmingly sad. What did this mean for Rylee's future? At the time I was in denial and unable to comprehend all the information about TSC. Luckily, my mom dealt with the diagnosis by doing a lot of research and compiling it into a huge three-ring binder for Rob and me. It was such a blessing that when we were ready to learn about the "scary" disorder known as TSC, the information was there.

After coming to grips with the diagnosis, we made a difficult decision. Because we learned that TSC was a genetic disorder, we decided to not have any more children. My picture-perfect life was slowly unraveling, and we had to accept the path our lives were going. We were only going to have one child rather than the two happy, healthy children I had dreamed about. But, once again, God had a different plan for our lives, a couple weeks after coming home from the hospital I learned that I was already pregnant.

Rob and I had mixed feeling about the news. It took us a couple of weeks to come to terms with Rylee's disorder and the news of the baby. When we were finally genuinely excited about the baby, I had a miscarriage. Once again I was crushed. Since neither Rob nor I had any obvious signs of TSC, it was determined Rylee's case was most likely a sporadic mutation and any other children we had would probably not have the disorder. The knowledge about TSC, along with the strong desire to have another child after the miscarriage, helped lead to my third pregnancy and our son Jake was born in December 2003.

Rylee was now two-and-a-half years old, and we had been living with the knowledge of the disorder for a year. We had a pretty good system for Rylee's medications and doctor appointments. Jake seemed like a happy and healthy baby, yet the fear of TSC was always in the back of my mind. Rylee's neurologist said to bring Jake in with Rylee, and he would monitor Jake along with Rylee. The problem was that I was in denial. I already had one child with TSC—God wouldn't give me two, would He?

I do not know how long I ignored Jake's white spots and staring spells before they got out of control. The day after Thanksgiving 2004, my 11-month-old son was having a seizure every 15 to 30 minutes. He was admitted to the Pediatric ICU at Loma Linda to get his seizures under control. Was this really the life God had planned for Rob and me and our children?

Rylee and Jake both have tubers in their brains that cause petite mal and complex partial seizures, but the rest of their internal organs seem healthy. They were both being followed by Inland Regional, an organization that helps children considered to be at risk developmentally, but they seemed to be right on track for their ages.

My children's TSC led to a diagnosis of the disorder in my husband, brother-in-law and father-in-law. Despite all the turmoil, time passing allowed life to slowly become manageable again. Rob and I were working parents with jobs that we loved. Rylee went to preschool, and my mom watched Jake during the day. Both Rylee and Jake were on different combinations of seizure medications and Rylee started a behavior modification medication, but life was good. The seizures seemed to be controlled and both kids were developing at a "normal" rate. I felt extremely fortunate. Rylee and Jake made me a better person and were a blessing to anyone who knew them.

When Jake turned two, Rob and I began discussing and praying about having a third child. Our two children were so incredible they made us want more. We discussed the option of adopting, but I had this overwhelming feeling of wanting to give birth to another child. Rob and I knew that some day Rylee and Jake would be faced with the same difficult decision and risks, and we wanted to set the example that life was worth it. We knew the percentages of having another child with TSC but believed the odds were in our favor. I honestly felt God would give us a “healthy” child free of TSC. Most people thought we were crazy, but we ignored the doubters and I soon became pregnant with Luke. The picture of my life had to include one “healthy” child, right? But once again God had another plan.

This time when I found out I was pregnant, I had an amniocentesis to check for the TSC mutation. Once again I felt my world come crashing in around me when I received the phone call that my new baby boy would also have the disorder. A fetal echo showed numerous growths in the heart. Soon after Luke was born, an MRI showed numerous tubers in his brain. Not only did Luke have TSC, but he also seemed to have the most clinically severe case of all our children.

Luke was born in December 2006 and spent 11 days in the NICU at Loma Linda Children's Hospital with SVTs (a heart problem that causes the heart rate to double) and possible seizures. The neurologist wanted Luke to take phenobarbital and vigabatrin (medication not yet approved by the FDA) for infantile spasms, and the cardiologist wanted him on medication to regulate his heart rate. When I heard this news I fought to keep back the tears, but I knew in my heart that it would be okay. It is funny how your brain tells you one thing, but your heart often tells you another. I realize the importance of listening to logic, but would not have my three amazing children if I had not listened to my heart.

My husband turned 30 last November, Rylee will be six this March, Jake turned three last December and our baby boy, Luke, is now two months old. TSC is a way of life for our family. People ask me if I wish I had a “normal” child. My question to them is to please define “normal?” Rob and I work every day to teach our children that having TSC is one of the many things that makes them so special.

Life is not easy, but having three children with TSC is an incredible blessing in disguise. When Rylee was diagnosed, I thought my world was coming to an end. I cried and cried for my baby girl and the difficult life she was going to have. Then, Jake came along and I cried, but had so much more hope. Rylee is an amazing example of strength and survival, and she passes that onto her little brother every day (including teaching him how to swallow pills.) When I learned Luke also had the disorder, it was extremely difficult to contain the tears, but I had no doubt that Luke was extremely fortunate to have Rylee and Jake for support as well as two parents who have already been through the diagnosis before.

I am a different person today than I was five years ago and experiencing TSC has made me a much better person. I am not going to say that life is easy, because it isn't. I am not going to say that my heart doesn't break with every hospitalization and complication, because it does. But life is worth it and my children give me hope every day for the future. I have learned to become extremely organized and patient. Looking back, not only am I a better person overall, but a much better mother, wife, daughter, friend and teacher. I have a higher level of compassion and understanding of those around me and am less judgmental of the human race in general.

When we decided to have a third child, the doubters told us we were being selfish and that it was too risky. In a way they were right. Rob and I were being selfish, because we believe that our lives are so much better with our children. We wanted Rylee and Jake to know that TSC didn't scare us; that no matter who you are or what type of disorder you might have, your life is special and impactful. We wanted them to know that they made us want more children just like them and if TSC was part of the package, it didn't matter.

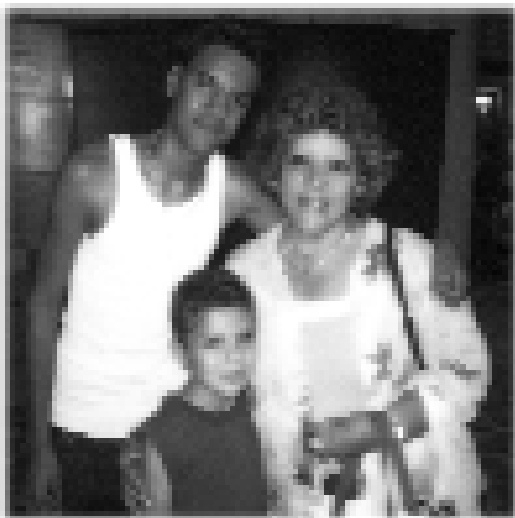
Our hope is that one day, Rylee, Jake and Luke will not have to ponder, pray and stress about the same decisions we had to make. Rob and I pray that research will find a way to manipulate the TSC genes, and that the seizures and TSC will not drive the decisions my children make for their futures. We are so grateful and blessed by the three miracles we've been given to raise.

Rylee, Jake and Luke are going to make a difference in this world. They have already impacted so many people in their short lives, and I have no doubt great things are in store for them. No, when I imagined my life I never dreamed about a really "scary" disorder known as TSC, but my life would not be complete without it. I now realize I do have the "picture-perfect" life, and it gets better every day!

Julio's Story

by Carmen Mary Alequin

On May 18, 1999, when Julio was born, I had great expectations for him. Julio is my fourth child. He was the only one delivered by C-section due to complications and 24 hours of drug-induced labor. I remember my first eyes on Julio. He was big and fat, blonde headed, with sky blue eyes and skin white as snow. Oh yes, he was heavenly sent. All I could do was thank God and cry. Julio was a good-looking baby. The only thing troubling me was that he had white marks and a small skin rash on his lower back. According to the doctors, this was of no concern and again said they were birthmarks.



Julio was a smart little boy. Julio easily graduated from Head Start to kindergarten. He was an eager little guy. Then at around the age of six, we started to notice he wasn't acting like himself. His homework was a struggle, his behavior didn't seem normal, and he became very argumentative. It was even to the point that he would make me and his older sister Joy cry due to his verbally abusive phases of rejection and hate, but they were words he didn't truly mean. To hear his little voice and see the face of an angel with such anger was devastating. But in minutes his lovable personality with hugs and kisses would heal the pain, and Julio wouldn't remember a thing.

At school even his teachers were starting to complain about some of the same things that I was noticing. At night, he was crying, screaming and jerking in bed. Sometimes hours would pass into the early morning. Julio would be playing and talking up a storm just about anything. When Julio would finally fall asleep it was time for us to get up for the next day's busy schedule of work and school. We became concerned and spoke to his doctor but Julio's actions were considered nightmares. The marks on his body (from birth) were still considered birthmarks, and they said the abnormal body movements during his outbursts were related to his behavior. These actions occurred in school as well, where they were handled as behavior problems. As a result, Julio didn't get to participate in recess or additional class activities. He was even sent to the principal's office, which terrified Julio.

On February 2, 2006 a call at work from Julio's school marked the beginning of our real nightmare. Julio had uncontrollable body movements, flapping of his arms, jumping up and down, and body jerking. For fear of the unknown, an ambulance was called. I was taken to the hospital by a co-worker, Mary Ann, to meet the paramedics as they rushed Julio into the hospital for proper care. At this time, I did not know that my son had just experienced a seizure.

A couple of tests and a scan of Julio's brain showed abnormal results. We were then referred to a neurologist for further testing. It felt as if every day brought a new doctor and more testing. Julio was to the point that he would scream that the doctors wanted to explode his brain. Julio would ask why I was permitting so many doctors to go into his head. It was so heartbreaking to try to explain to a six year old that something was not going right in his little head. He would ask me why God made him like this.

After almost a full month of testing, the appointment came for us to get the results. We will never forget the date of March 1, 2006—the day our lives were changed forever. Julio was diagnosed with tuberous sclerosis complex (TSC). What? TSC, what dear Lord is this? I sat there in shock, and tears rolled down my face uncontrollably. I began to shake. I felt nauseated and I wanted to faint. I asked why us? What did I do wrong? Why my beautiful baby Julio? Why? Why?

As the doctors tried to comfort me, Julio was scared and also began to cry. “Mommy, am I going to die? Why am I turning different? Mommy what is the doctor telling you?” he screamed. I then began to turn to my faith for strength. As I began to calm down with Julio in my arms, the doctors just let us hug. My spirituality helped me get through that day.

It's just always been the four children and me to face whatever challenge came before us. Now it was time to face our worst fear because of Julio's diagnosis. As the doctor continued to speak, every word was a sword through my heart. He said TSC is a disorder that causes tumors to form in various organs, primarily the brain, eyes, heart, kidneys, liver and lungs. People with TSC often develop epilepsy, autism and learning and behavioral problems. But the sword that stays plugged in our hearts is from the doctor's last words—*currently, there is no cure*.

As if this wasn't stressful enough, we faced the hardship of multiple doctor appointments to determine which medication and dosage would be best for Julio to control his seizures. Upon taking the medication, Julio had problems digesting it. He was also experiencing numerous seizures and mood swings. School revealed that he was losing his motor skills and becoming cognitively delayed. And, his behavior had to be monitored.

All this was happening to my Julio and having to deal with the ignorance of the school system is an unbelievable tragedy in itself. The school system is not familiar with this disorder and I am frequently challenged about Julio's needs. I am constantly being called about Julio's behavior. Even on things that are known to the school (because of all the information that I've given them), I am still questioned.

At one point it was called to my attention that maybe Julio should be transferred. But a co-worker and a special education counselor looked into the laws to protect medically ill children. As a result, a 504 plan was put in place immediately. I have become Julio's advocate by reading, studying, asking for handouts, searching the Internet and attending classes whenever possible. I fear for Julio's education. I want him to get as much education as he can absorb.

My concern is when the cure is found for TSC that Julio and thousands of others will not have the proper education needed for them to continue to function in this world. Does this mean that Julio and others will not be able to do anything productive because he was never helped from the beginning?

There is so much to say but so little time, for every minute counts in the lives of our loved ones with TSC. I will just give one example of time and how fast TSC moves inside the body sometimes—Julio has not been diagnosed more than a year and his vision has gotten worse, so much so that he must wear glasses. He has a hard time seeing.

Julio has also been diagnosed with Attention Deficit Hyperactivity Disorder (AD/HD). Now I must go back to the school to amend his individual education plan so that it meets the needs that Julio now faces. Once again, I will deal the stress, hardship and tears in dealing with the school system that doesn't understand.

My family knows this is just the beginning because TSC affects so many aspects of life—the physical, the mental, the hardship of finding the right medical help, identifying proper education needs, and even obtaining social security (which I have been denied for Julio). And I could go on.

Although I know better, Julio feels as if he can walk anywhere and nothing will hurt him. He doesn't fear the fire on the stove. If Julio wants something from the skillet in the back burner, he will just go over the other skillet to take whatever. Julio takes everything you say to heart. At school, a teacher told the children not to come to her desk unless there was a fire or if they were bleeding. Julio took it literally and tried to bleed by scratching his arm with a pencil so the teacher would let him talk. I addressed this matter as I have with a couple of other incidents.

When Julio has a light or small seizure, people think he is a spoiled kid having a tantrum, and remarks are always being made around us. If Julio is corrected for something that he has done, he will have a tantrum. When he calms down, he does not remember or even realize what he has just experienced. He will smile or just walk away, minding his own business as if nothing ever happened. If Julio has a question, he will repeat it rapidly. These along with a thousand other things are very upsetting and frustrating. So you see this affects the whole family.

TSC has also changed the lives of my other children, especially on an emotional level. Because Julio is changing in so many ways, the other children are affected and devastated and they cry about not knowing what the future holds for their baby brother. Will Julio be able to understand and know how much we all love him or will he not remember in time?

There is so much for all of us to handle. As for myself, I've been so overwhelmed with all of this. As the sole breadwinner, I get so stressed out that I don't take care of myself and I almost ended in the hospital. I am now under doctor's care for stage two diabetes.

So you see, TSC isn't just in the life of my son Julio, but in the lives of all those who care for him. We must move forward for we are fighting the hands of time. Let's make a difference today.

Julio also is doing his part and learning to advocate in moving forward for TSC. Julio has spoken to doctors at a conference where the doctors themselves were unaware of the disorder. Julio was a little model for the doctors who had not seen the clinical features of TSC.

Little Julio also visited with State Senator William Delgado to tell him how he wants and needs help with TSC. Mr. Delgado told Julio that he would help. Julio then received a call from the Illinois State Medical Society on behalf of the Senator. Let's see what the future holds.

Julio also goes to most support group meetings with me. Some days are harder than others for Julio, but there he sits asking questions. Of course, sometimes he gets up and moves around, but at least Julio is making a difference. I will do whatever I can to help Julio reach his full potential.

In the meantime, we are enjoying every minute we can with Julio. Julio's favorite pastime is horses, and he loves to ride them! His godfather Wo, as Julio calls him, tries to take Julio horseback riding in Wisconsin as much as he can. Julio is a very happy little boy. He loves life and sings to the Lord as often as possible, especially when in Wisconsin. Julio has a good heart and that's what we'll always keep close to our hearts. I still cry when I talk about Julio and his diagnosis, but I can't wait to cry for joy when there is a cure.

Ethan's Story—Update

by April Bennett

Ethan, the light of our lives, arrived on May 11, 1995. David and I eagerly welcomed his birth. We spent the entire nine months anxiously preparing for him by attending to every detail, his room and every possible baby item sat at our house ready to put into service. Little did we know that we would only have 51 weeks of a “normal” family experience. I will forever be grateful for those 51 weeks.



On May 3, 1996, our world came crashing down on us. During a routine annual check-up, Ethan was diagnosed with tuberous sclerosis complex (TSC). Within minutes, the words mental retardation, seizures, tumors, learning and behavior disorders came flooding over us in reference to our precious baby boy. Time felt like it was slowing down. I remember looking at my husband who held our son sleeping on his lap. He had his arms wrapped around Ethan sobbing. Looking back at the doctor I heard her rambling on and on with symptoms of this disease. I wanted to scream, “That’s enough, Stop!” How could this one disorder cause so many symptoms?

Ethan had been experiencing a type of seizure called infantile spasms. As it turns out, they are the worst kind. After an appointment with a pediatric neurologist, I administered the injection of ACTH once a day. Until that point, giving Ethan a shot every day was the toughest thing I ever had to do. I didn’t have to do it; I chose to do it. With every ounce of preparation for Ethan’s arrival, nothing could have prepared us for TSC. As Ethan’s mom, I felt responsible, and I had to force myself to give every shot.

As far as I am concerned, David had the hardest job. David had the responsibility of holding Ethan’s hands and keeping him still for each shot. He had to look into Ethan’s big brown eyes. In his eyes, David could see a child who didn’t understand why we had to do this to him. We both knew it had to be done. Living with TSC, we constantly have decisions to make where logic versus emotions makes it very difficult.

After 45 days and 35 shots, Ethan finished his ACTH treatment. He had experienced one mild seizure approximately 12 hours after the first shot. Since then, he had been seizure free. Every day afterward, we took one day at a time trying never to jinx it.

After getting the seizures to stop, we witnessed an instant leap forward in his development. By 17 months, Ethan was caught up in every area except speech. This would continue to be an area where early intervention focused until his third birthday. Everyone kept trying to reassure me that boys never talk early. I tried to reason with myself, “Ethan is an only

child, and I meet his needs and he doesn't have a reason to talk." The entire time, Ethan was probably snickering at me knowing he could talk and just waiting until the right time to prove it.

A few months after his second birthday, David and I got the best present ever: Ethan suddenly could say anything he wanted! Almost instantaneously, he began stringing together words. This was a joyous time in our lives as Ethan was seizure and medication free, and his development was on target.

Turning three meant that we had to transition from early intervention to the early childhood program within the school district. Shortly after his third birthday, he was evaluated for this program. During the fine motor skills section the psychometrist asked him to touch his fingers together, and Ethan blurted out "Dr. Pepper!" This confused the woman, so she asked us why he said this. We told her that the finger movement that she asked him to do looked like the "sign" for Dr. Pepper. She burst into laughter, and admired his intellectual ability. Ethan failed to qualify for early childhood services with their label that "he is too smart." This news didn't disturb me. I reveled in the fact that my son with TSC was "too smart" for their program.

Ethan and I enjoyed spending our days together instead of going to school. Pre-K, the program for four year olds, would be where Ethan would start his schooling. Dropping him off that first day was unbearable for me. Ethan could have cared less. Entering his classroom, he turned around and said "Bye Mom." I managed to leave, but not without tears rolling down my face. Starting Kindergarten mirrored the first day of Pre-K. Ethan was excited to be around all the other kids. Probably he was also ready for a break from Mom.

First grade brought a bit of anxiety for us both. This was the first year Ethan would be in the school building, not a mobile unit outside. We weren't sure he could handle this transition. During this school year, Ethan had problems completing assignments. Apparently, he didn't want to stop talking to his neighbor to do his work. I am thankful that his teacher gave him some flexibility. Instead of making first grade a horrible experience for him, she allowed him to take the work home and finish it.

Second grade brought other issues. Signs of attention deficit disorder (ADD) began to rise to the surface, so we had Ethan tested and he scored just below the level of diagnosis for ADD. During this testing, he was diagnosed with anxiety issues. David and I knew that this was an issue, but with counseling Ethan was able to overcome these problems for several years. Third, fourth and fifth grades blur together for me. Ethan continued to perform without a need for special education services. Ethan excelled in language arts but struggled with math. Throughout his elementary years, he remained medication and seizure free.

Transitioning into the sixth grade appeared to create panic attacks. In August 2006, Ethan began to tell us about "episodes" he had experienced. We questioned Ethan, trying to determine what these episodes could be. To the best of his ability, he claimed they started with a nervous feeling. The appearance of these episodes and the timing of heading into the sixth grade kept us focused on them being anxiety related. To be safe, we went in for an EEG, which came back normal. David and I felt relieved and moved forward with medication and counseling. With this difficulty, the school district placed him on a 504 plan, which means he gets some accommodations and modifications based on his medical status.

Valentine's Day, February 14, 2007, is another day we will never forget. While getting ready for school, Ethan experienced an "episode." After 10 to 15 minutes, I called the doctor and rushed him to the emergency room. Ethan went into a status seizure that lasted close to three hours. Dose after dose of Ativan® failed to stop the seizing. After about an hour, the seizure moved into a complete partial type, starting in his right side. We saw one finger twitching, then all of them, then his arm, then his leg until his entire body jerked and thrusted; we had to roll him on his side to keep his airway clear. Finally, Cerebyx® was administered through his IV, and he began to relax and transition from a seizure state to sleep. He remained asleep off and on for the rest of the day. Watching my son's body move about without him being "present" horrified me beyond belief.

Somehow, through it all, I have found the strength to stay by my son's side during every test, stitch, seizure, shot, and more. That is my job. As Ethan's mom, I take full responsibility for the good and the bad.

Although Ethan was seizure free and medication free for more than a decade, David and I knew that it could all change any day. Educating ourselves about every aspect of TSC, and learning about experiences from other parents, is what saved Ethan that day.

There hasn't been a day since May 3, 1996 that TSC hasn't been in my thoughts and vocabulary. Watching my son experience normalcy in life didn't deaden my sense of responsibility.

A Bond Beyond Words

By Mark Martin

I remember when my wife, Nadia, called me on New Year's Eve 2001. I was working my regular shift and was just told I would have to work the next shift too. Nadia called to tell me that she thought our one-year-old son Alex had what looked like a seizure in his swing at home.

I remember the shock that hit me when those words were spoken. Could it be that Alex had a seizure? That was probably one of the longest nights of my life. As I anxiously awaited the shift to come to an end, I remember speeding home to see for myself what was truly occurring. But within a few short days, it happened: Alex had several more seizures and from that point on, my world changed. Not only because Alex had seizures, but the hidden answers and riddles that had always plagued me were finally about to be answered and come to light.



The diagnosis of tuberous sclerosis complex (TSC) was not only bewildering, but I could hardly say “tuberous sclerosis complex.” Those words marked the beginning of a medical and emotional roller coaster ride for not only Alex and our family, but for me too. When the doctor told me about TSC and the fact that it is a genetic disorder, I can't put into words the guilt that came over me.

We believe my father had TSC. I am an only child, and my dad remarried and had three more children. My sister Adella has TSC, along with her third daughter. That was how the TSC finger points straight at my father. At first, dad was in denial and became quite stubborn about accepting the fact that he passed TSC to us. But when Alex had a few seizures in front of him, his mind changed for the most part.

I started having seizures when I was 12 years old. I had only nocturnal seizures and at first onset, I had about 20 in a row. After being told I was epileptic, I was sent home to begin taking Dilantin® and phenobarbital. Shortly after coming home and going to bed, I had around 20 more seizures. I was hospitalized and put into a net bed. My life was about to take a sudden and drastic turn. I was not allowed to play sports any more, and I remember my social life at school changed. I turned into an introverted, fearful social outcast. I remember life being full of dark hallways as well as being misunderstood and labeled for years to come. To this day, those cold, dark feelings and memories remain.

Two weeks after Alex's diagnosis, I developed the first of five blood clots. While I was in the hospital, I underwent a series of tests that determined I had TSC. My mind went into a lockdown of guilt and shock. Denial was not an option, because for the next year or so, Alex had a ton of what seemed like never-ending seizures. And there was even a time when Alex had a status seizure that lasted two-and-a-half hours, causing him to be placed on life support for a night.

But this dark cloud of TSC held its own silver lining for me. As Alex started to receive services like occupational and speech therapies, I started to get answers as to why I was the way I was all those years and who I was. I learned about sensory issues and more about the many side effects stemming from TSC. A new chapter in my life was opening up for me one day at a time. I was learning more and more about me because of Alex. My love for my son took on a deeper bond than I could ever have imagined. From the physical to the psychological sides of TSC, I gained more information in this short time than I had in 40-plus years of looking for answers. As if it was yesterday, I recall the feeling I had when my doctors would make me feel as if I were some kind of freak who had nothing better to do than to waste their golf time. I crawled into a hole of insecurity that took years to overcome.

Alex and I have a bond that goes beyond words. I know what he's feeling when he gets excited or upset. I know how he enjoys brushing and other sensory implementation, along with the satisfaction of deep pressure. I know how crowds and loud noises can be overbearing for him. I also know that Alex will always encounter those who do not understand him or his TSC—even doctors who claim to have insight to TSC but who really don't have a clue as to what it is and how it runs its course. I don't know if Alex will ever know the love and gratefulness that I have for and toward him. I don't know where I would be today without his portion in my life. He truly is my biggest hero. Without him, my life would be an endless walk of unanswered questions.

It has been a tough road of discovery for Alex and me. We moved to Tennessee last August and it sometimes feels like we have to start all over again. Yet when I met Dr. Kevin Ess by way of Dr Elizabeth Thiele, I knew we were going in the right direction of hope. I became active in the TS Alliance of Middle Tennessee, attended a few Regional TSC Conferences and have been to Washington, DC to speak to Congressional members about TSC research. I also attended the National TSC Conference in Chicago as an adult with TSC, as well as a parent of a child with TSC.

I have had the awesome benefit of getting to know my other family—all the wonderful people at the TS Alliance and all those wonderful friends I have met throughout the last five years. I have had the great and moving experience of working on the TS Alliance's Adult Issues Committee along with another awesome friend, Nikki Seefeldt. Nikki has been a source of great inspiration and a faithful friend. I would not have been put in this position if it wasn't for Nikki's faith and guidance in and toward me. It's funny how a disorder like TSC could have a person like me in bondage without knowledge to help set me free. But even more amazing is that my son ended up holding all the keys that would unlock so many solidly shut doors of my past.

I did not start talking until I was four years old. Alex is seven now and is still non-verbal. But I believe with all my heart that Alex will talk despite his age and despite those who said he probably won't. Alex and I may have TSC, but TSC does *not* have us, for we have an unbreakable bond of love that TSC will never penetrate nor take apart.

I have seen many doctors, and I have been very frustrated at many times in my life. Even after our discovery of TSC, the road appeared to become narrower and distant, not knowing who to trust or believe. I often wondered what was worse: Not knowing anything at all, or knowing something and yet being pushed aside with the status quo medical answer and opinion. In many ways this is a heart-wrenching story to share. But I share our story because my focus is on leading the way for my little Alex, to teach him that no one can ever keep you from doing your best to reach for your own answers and hope for a cure.

A Tale of Woe and Hope

by Herbert Zwickel

Our daughter arrived in a quick and furious manner. At birth it was determined that our daughter Katrina was having respiratory problems. Instead of coming home she was taken to Westchester Medical Center, a large facility near New York City. Her arrival home was delayed by two weeks. An answer was never given as to what caused the problem. Katrina was a beautiful baby. We began to notice a red mark that was raised and in the shape of a heart under her left eye. We were informed that this was a birthmark and could be easily corrected when Katrina got older. We were happy to see our daughter come home and settle into a normal lifestyle. We accepted the diagnosis of various doctors concerning our daughter's health and felt satisfied that everything would be okay.



Katrina was a very bright baby and met all of her milestones early. As time went on, we noticed certain behaviors, which were a concern to us. At the age of four Katrina began to sleep walk periodically. We realized its seriousness when she was able to sleep walk right out of the house. We addressed this with her doctors, who said she would out grow it. At around the age of seven, we began to notice non-pigmented spots on her skin. At this time, we saw a team of doctors who were interested in her peculiar markings. They did a full examination and encouraged us to have her birthmark removed with laser treatments, but the treatments were unsuccessful. They also did two biopsies of her birthmark to classify it and determine whether it was cancerous.

At age twelve she developed a scarred area on her lower back. The sleep walking was becoming a nightly situation. We were naturally interested in the fact that the birthmark, which doctors said would go away, had not. At the age of fourteen Katrina became very withdrawn and refused to be left home without a parent. At night we would hear sounds emanating from Katrina's room. These sounds were a mixture of her grinding her teeth and moving in her sleep. She did not respond when we tried to wake her. With these issues coming to the forefront, we began seeking medical help once again. We saw a neurologist in New Paltz, New York who entertained our concerns. He prescribed a three-day, at-home EEG. When he reviewed the testing, he informed us that Katrina was having partial-complex seizures and prescribed Trileptal®. This did not sit well with us because we were still unsure what caused the seizures, so we did not give her the medicine. We immediately made an appointment at the Neurological Institute of New York at Columbia University Medical Center. In November 2004, the day before our scheduled appointment, we were watching television as a family when we experienced Katrina's first daytime seizure. It was very frightening for all of us.

We arrived anxiously at our neurology appointment the next day. Much to our surprise, Dr. Cigdem Inan Akman told us that based solely on her physical examination of Katrina, she was 99 percent certain our daughter has a disease called tuberous sclerosis complex (TSC), but needed a brain MRI to confirm the diagnosis. This was a tremendous breakthrough for us; Katrina's condition could now be identified. We went from not knowing anything to knowing what her disease was. Katrina began taking Trileptal, and her nighttime activities ceased.

We have been told that TSC can affect a person in a negative way as far as academics and mental ability. Katrina is, and has always been, an excellent student. She was placed on high honor roll in both junior and senior high school. She has also been in the National Junior Honor Society and is currently in the National Honor Society. She is a junior in high school and will begin her senior year in 2007. As a result of her academic ability, she has been invited to participate in numerous exciting activities. At the time, we had no knowledge of her real medical problem, so we were forced to turn them down.

We were asked to have blood tests to see if the TSC was a genetic problem within the family. The testing showed that Katrina had a genetic mutation, which will only affect her and her offspring. We now know the beginning of the road that our Katrina has to walk down. The more we found out about TSC, the more frightening the situation became. One difficult aspect of TSC is not knowing other folks within our community with similar problems.

After Katrina was diagnosed with TSC, she began to face another issue. Three of her lifelong friends began to walk away from her. We believe that Katrina's medical issues were so obvious to them that they lost all interest in her. Things got so difficult for Katrina that we had to deal with the high school to help us separate her from these people that used to be her best friends. This was a most difficult situation and took many months of patience and understanding for Katrina to realize that nothing could be done to change their minds. Katrina did have a few friends who stood beside her the whole time. They saw beyond Katrina's illness, and loved her just the same, maybe even more. Katrina was finally able to move on and began to make new friends who liked her for who she was. In time, Katrina found new outlets for herself. She studies and teaches dance, plays the baritone horn in the wind ensemble at school, studies piano and is doing college credit work in high school. This summer Katrina will be working on a project dealing with TSC.

We realize that the future can be most uncertain especially for people with rare diseases. We love our daughter and we trust that, Lord willing, she will have a good and productive life. Her desire is to become a medical doctor. We know that this is a hard task and a difficult road to follow, but we trust that she will succeed in her efforts and hopefully contribute to the field of TSC. We have the hope that we can win in this difficult medical war and that our daughter will be able to add to the knowledge required to gain the victory.

It is rather obvious that at this time there is an organization, the TS Alliance, that helps folks with TSC and their needs. I know it was important for our family to meet others with this problem and to realize that this disease afflicts the body in various ways. It is our hope that we can help others through marches, rallies and outright contributions. There is certainly a need to contribute to the research, to petition federal and state governments for research and to help those who cannot afford the high medical costs. We trust that all our TSC families would be involved with good TSC programs that seek answers to find a cure.

My TSC Journey

By Julie Fleming

I did not know I had tuberous sclerosis complex (TSC) until I was about 20 years old. About 10 more years passed before I started to understand the ramifications of this diagnosis. As a child, people always asked about my unique, large white “birthmark” on the back of my right leg. It was just always there. This mark never bothered me.



During adolescence, small red bumps began to form in a small line across my nose. A small bump began to grow on the nail of my littlest toe. These bumps annoyed me, especially as a self-conscious teenager, but I was not too worried. My dad had similar bumps on his toes, fingers, and across his nose, cheeks and chin. My siblings also developed similar bumps. I figured it was some genetic trait from my father like bad eyesight or thick hair. Little did I know...

My story is intertwined with my father's story. I need to share part of his story before I can continue. When my father was young, he had mysterious red bumps appear on his face, and bumps grew on his fingers and toes. His mother was baffled and did not know what these bumps meant. Doctors were perplexed and made many erroneous diagnoses, including bad acne. None of their diagnoses were correct. When my father was in his 30s, he had cancer on his kidney, and the cancerous kidney was removed. The doctor mentioned TSC as a possibility, but felt this could not be right because my father had no cognitive impairment, intellectual disability, seizures, or behavioral issues.

Finally, after losing one kidney to cancer, going through years of renal failure due to angiomyolipoma growth on his remaining kidney, and having unexplained red bumps and growths on his face, fingers and toes, my father was officially diagnosed with TSC when he was about 48 years old. His doctors were amazed that he was a high-functioning adult with TSC. They were even more shocked to realize that he had three children who were also high-functioning TSC adults. My father was put on dialysis and later received a successful kidney transplant. His second kidney was then removed due to the excessive tumor growth, which greatly reduced his kidney function.

At this time, I was about 20 years old and in college. My siblings and I knew we must have this same disease because of the skin manifestations we had in common. In my mind, this meant two things for me. One, I could some day develop kidney problems. Two, I could pass this disease to my children.

After I was married, I went to a dermatologist to have my many moles and other bumps checked out. The doctor assured me that I did not have TSC, because I only had the characteristics on the skin, not the disease itself. This assessment gave me a brief glimmer of hope. I thought he could be right. He was the doctor after all and shouldn't I trust the expert? Reality then set back in, and I knew he was wrong.

When my brother was about 24 years old, he started having severe headaches. The doctors discovered a large apricot-sized subependymal giant cell tumor (SGCT) on his brain, which was surgically removed. Seeing him in the hospital after the surgery was especially difficult because I knew that could be me someday. However, I assumed it would only be a problem if I started having bad headaches. I am not sure if I was in denial or truly did not understand that I should be having my brain checked for tumors.

When my husband and I decided to have children, we knew our chances were 50/50. It was a risk we decided to take. We had a 50 percent chance that our children would inherit TSC—not bad odds. Plus, the doctors told my father and brother that our family has a “mild” case and we were lucky. I told my obstetricians about my genetic disease. At the ultrasounds, we asked them to look closely, knowing there was a possibility of tumors growing on the internal organs. Nothing was ever found. My son was born in 1995 and had no TSC-related problems.

Two and a half years later, my daughter was born. Within 24 hours of her birth, the doctor heard an irregularity while listening to her heart. She was only 4 days old when we took her to Primary Children's Medical Center for a battery of tests to discover the cause of this irregularity. The reality of the bad 50/50 odds was staring me straight in the face. Taking her from test to test became unbearable. I was consumed with worry about what the future held or did not hold for her.

After many tests and long hours of waiting, a cardiologist confirmed the diagnosis. She had multiple rhabdomyomas on her heart that were blocking the flow of blood as it exited her tiny heart. She was the recipient of my bad genetics and had TSC. I felt incredible amounts of guilt for passing this on to her. I shed many tears grieving over what I gave her. I have never felt more helpless.

I remember coming home and researching rhabdomyomas. I found out everything I could. I was so incredibly grateful to discover that these tumors tend to shrink or dissolve over time. As her heart would grow, these tumors would not. Thankfully, she had the only kind of TSC tumor that doesn't grow. I felt a sense of relief. I know it could have been so much worse. Over time, most of my daughter's rhabdomyomas have completely dissolved or become so small they are insignificant.

For a while, we had to take her to get routine echocardiograms to monitor these tumors. She had to be sedated, which was a nightmare because she would fight the sedation and was not herself when under the influence of these drugs. It was hard to help her. During one visit, the cardiologist decided to check with a geneticist about other tests. The geneticist recommended tests to check her brain and kidneys. This was the first time a doctor recommended any additional testing. She had these scans and she was clear.

The geneticist further recommended that my son be seen by him to rule out TSC. We took my son to see the geneticist, who used a Wood's Lamp and found nothing. He then put the lamp on my leg and arm. My ash leaf and confetti spots lit up like a Christmas tree, so the geneticist recommended that I have scans of my lungs, kidneys and brain. This was the first time a doctor ever told me to get checked. I was 30 years old. He was sure I had TSC. He was only the second doctor to ever say that to me.

I finally went to my regular doctor and arranged for testing. Through these tests, I learned that I have three "nodules" growing in my lungs. I have also developed pitting in my teeth. I have a small white patch of hair that is TSC-related. I have almost every skin manifestation of TSC, but my brain and kidneys are clear so far.

Combined, my brother, sister, and I have eight children. Five of them, all girls, have TSC. All of the girls have hypopigmented macules (white spots on their skin). Some have bumps on their toes and faces. One niece takes medication to control her mild seizures. She has small tubers and a small SGCT growing on her brain.

Over the years, I have had to tell the doctors what my daughter and I have and educate them about this disease. At times, this is frustrating. I have learned that knowledge is power. I need to know what the best options are for my daughter and me. The more information I have, the more respect I am given in return.

My family has what are considered "mild" cases, yet every organ of the body has been affected within our family with one exception, the eyes. This disease is unpredictable and a struggle no matter how mild or severe it may be.

In recent years, I have become much more proactive in fighting TSC. My greatest hope is that some day researchers will find a cure and my daughter can be free of TSC. I truly hope that some day, when the time comes for my daughter to have children, that she has a zero percent chance of passing TSC on to them.

Reach for Hope

By Gwen Pryor

I was born on October 22, 1963. This was about 10 years before the National Tuberous Sclerosis Association was formed (the name the TS Alliance held in its inception). My mom recalls that at about five months of age, she noticed my head would drop very oddly while I was in my crib. This would continue day after day. The doctors really dismissed her concerns at that time, until at about 19 months, when I began having what today are called tonic clonic seizures. My parents were frantic. I was their first child and no one seemed to know what was going on. At that time, the premier drugs were only phenobarbital and Dilantin®. Both of these drugs failed.

Unsure of what to do, my parents traveled between Chicago and Detroit and to our local hospital, featuring my case at any symposium they could just to find an answer about my seizures. During this time, they also noticed I had the white patches of skin (now called hypopigmented macules) on my legs. These trips lasted almost two years.

When I was about three, I started developing pubic hair and breast buds. The doctors then went in the direction that possibly I had a tumor on my pituitary gland and gave my parents the option of exploratory brain surgery to find out what was causing this. They flatly rejected this approach. All the while, I continued to have seizures. My mother talks about the time I went into the hospital to be sedated to have a pelvic exam at age three. The doctors couldn't get me sedated from all of the medications I was on, so when I finally went "under," the doctors couldn't get me out of the sedated state so they started slapping me just to get me to come to.

Finally, at almost age four, my parents were able to obtain an appointment with a pediatric neurologist who only worked on convulsive disorders in children. This doctor commuted between Chicago and London and agreed to take on my case. After almost two years of seeking a diagnosis, he looked at me, without touching me, and told my parents he thought he knew what the problem was: tuberous sclerosis complex (TSC). My parents were amazed. After all of the testing done to no avail, this doctor could diagnose me with no problem! He noticed a small growth in the crease between my cheek and nose, on the left side of my face. Dr. Millichap felt that if they did a biopsy of the growth it would be found to be an angiofibroma, a characteristic of TSC.

That biopsy would prove to be the conclusive diagnosis of TSC. But my seizures still continued. At that time in 1967, I was put on an experimental drug here in the United States called Mogadon®. To my understanding, this drug still hasn't been patented here in the United States due to some really severe side effects. It is currently being used in the UK and other countries as a sleeping pill. It has also recently been put back on the epileptic drug list. So it may be patented again in the future. Anyway, I was put on Mogadon and my seizures stopped. I was about 4. My mom, along with the help of our local pediatrician, was able to wean me down to no medications, and to this day at 39 years of age, I have been seizure-free and medication-free.

I went to school, but had developmental delays. The doctors fought the school system to have my case classified as a medical disability versus a learning one. But I soon caught up to my class. I went through speech therapy in second grade; otherwise I grew up in a mainstream, private Christian school classroom.

By middle school, I was earning As and Bs but was painfully shy and had few friends. That could be the topic of another story, but suffice it to say that kids in middle school are very cruel toward kids who are different. Add to the fact that my angiofibromas had become very pronounced, I was painfully shy, and I had indeed gone through puberty early, my self-esteem was at an all-time low. As a result, between junior high and high school when I was 13, I underwent total facial dermabrasion at the University of Michigan, which is a couple of hours away from my house. Today, many children who undergo facial surgery for their angiofibromas have laser surgery, which is a lot less invasive and a lot less traumatic with very little pain afterward. With my surgery, my whole face was done, and I was in the hospital for an entire week. At that time, dermabrasion was experimental for TSC, and the doctors really didn't know how the outcome would be. It has been 30 years and I have yet to have the procedure repeated. With a bit of makeup, the remaining angiofibromas are hardly visible on my face.

I went back to school in the fall, and finished high school with no medical issues. All the while, I did not know anything about other issues related to TSC. I dated and went through all of the high school things. I ran cross-country track, played in the school band, sang in the choir, and appeared in the school musical one year. I wound up earning a scholarship to college.

After high school, I went on to college and earned a bachelor's degree in accounting. Six months after graduation, I met my husband, Dave. I still hadn't had a TSC checkup since my seizures had ceased. But a pediatric neurologist who was a customer of the bank where I worked introduced me to the National Tuberous Sclerosis Association (now TS Alliance). At that time, I was in great denial that more could be wrong with me. After reading all of the TSC-related literature, Dave and I opted for genetic counseling about our future family. We had been married about three years at this time. The counselor confirmed the TSC diagnosis and marveled at how well I was doing. Based on all of the genetic information, we decided to adopt rather than have biological children.

I will mention what another parent told me about having children with a TSC diagnosis versus not, which I felt was very powerful. She told me that we both live with losses. There is a one-in-two chance of passing on TSC...1 in 2! Would you put your house, marriage, everything you hold dear on a one-in-two chance? Somehow, 50-50 sounds better doesn't it? While we made the conscious choice to not have biological children and that was a huge loss in our lives, she chose to get pregnant and had a child with a spontaneous mutation of TSC. I will never carry a baby in my body, breastfeed, or experience birth. While she did all those things, she has the loss of having to become her son's guardian. He will never live independently, will never marry, and will never make her a grandma. Her question to me was, "What loss can you handle more, especially in the world where a TSC-affected adult has a one-in-two chance in passing it on?"

Also, I was once told that TSC splits in thirds: one third is mildly affected like me, one third has some issues, and one third is severely affected. A big issue for me is that while I am against abortion and personally have a moral problem with embryo selection, my

Lymphangioleiomyomatosis (LAM) diagnosis complicates things. So my husband and I had to ask the question: Could we look into our child's eyes without guilt if we conceived a severely affected child? That question has a different answer for everyone—for us the answer was that we could not go down that road no matter how difficult it has been.

Following the adoption of our oldest daughter, I had my family doctor do an ultrasound of my kidneys to convince me that there wasn't any kidney involvement. The ultrasound came back with masses on both kidneys. I was devastated.

My brother had had a different type of surgery (which had nothing to do with my TSC) that required treatment at Mayo Clinic. While my mom was with him for his surgery, she read all about Dr. Manuel Gomez, who was considered to be the father of TSC. At her urging, I went to Mayo in 1995 for my first checkup since about 1967.

The doctors found angiomyolipomas on both kidneys, a liver tumor, hamartomas on the back of both eyes, ungual growths (minor nail bed growths), hypopigmented macules, and the remaining angiofibromas on my face—all of which amazingly had been stable. In 1999, I was also diagnosed with another rare disease I mentioned earlier: LAM, a lung disease that can be frighteningly progressive in women. It is thought that up to 40% of women with TSC will be diagnosed with LAM. Diagnosis is done with a high resolution CT scan, usually during the childbearing years (between the ages of 24-35). Some women with LAM do get worse; however, LAM research is ongoing, and I have been stable on that front also since my diagnosis.

Today, I have been married 17 years, and my husband and I have three adopted children: Kelsey age 11, Nathan age 9, and Kimmie age 9. The job I had when we adopted Nathan was eliminated so I have been a stay-at-home mom since then. I have completed a course to do medical transcription and work full time from my home.

I live a totally normal life like anyone else. The only difference is that I go for a checkup once a year at the TSC Clinic in Cincinnati, OH. One thing I found interesting though: When my husband and I got all of my records from the pediatrician to go to Mayo back in 1995, we found a letter from the diagnosing neurologist to the family doctor. The last line really stands out for me because it said, "Her prognosis at best is poor." I recently found out that the diagnosing doctor is still alive and would love to show him how my life has turned out. My mom was shocked to say the least when she read that letter and would later say she was glad she didn't know of its existence, because she felt it would have colored how my parents raised me. They didn't know TSC was genetic and all of the implications it held. My parents raised me to live in the "real" world and not place myself in a disabled world unless there was absolutely no other option.

I urge you, at this fragile time, to reach for hope. I know parents are scared. My advice is as follows: Doctors know more than they did in 1967; however, they are still proven wrong every day. Be your child's advocate. Let him/her reach for the stars in his/her life. Don't show defeat or he or she will grow up feeling defeated. I understand there are some serious issues with TSC, which is why I get checkups every year. Handle the medical issues, but realize that the life you have been given is a gift, whether you are an adult with TSC or a parent of a child with TSC.

Sissy's Story

by Gloria Schultz

Our journey with tuberous sclerosis complex (TSC) begins just before my second daughter, Angela, was born. It was spring 1987, and I was just 21 years old, living in Arizona. My husband was just 26 when his mom, Colleen, developed heart problems. She had no clue what her doctor was talking about when he asked if she knew she had TSC. She had the facial angiofibromas, a shagreen patch on her back, and lesions on her heart. One of Colleen's three siblings also had "the family skin," as well as two of her four kids, who both had a history of seizures too. Three of Colleen's kids had growths on their fingers and toes called periungual and subungual fibromas. One of those boys was my husband and my daughters' father, Tony. None of us had ever heard of TSC. Now it dominates our lives.



Angela, better known as Sissy, was born July 15, 1987. She was perfectly healthy and developed normally. A white spot appeared on her hand when she was 9 months old. I took the information I had about TSC with me as I went to two doctors that knew nothing at all about TSC. Then we were referred to Dr. Michael Frost at Phoenix Children's Hospital. He used the Wood's Lamp to confirm that the hypopigmented area and the confetti markings on her skin were indeed caused by TSC. He ran a CT scan on her brain and found tubers there. I remember being told that because TSC varies in all people with the disorder, that this may be the only sign we ever see and to not worry about things that haven't happened. We were also told how to recognize a seizure just in case it happened. Then it did.

My baby was just 18 months old and she loved books. She slept with them at night and cuddled dolls too. She walked on time, talked on time, and she went to Sunday school too. Then, over New Year's weekend she experienced unusual facial twitching and we knew she was having a seizure. It was the first seizure I had ever seen and it was in my own daughter. She recovered well and had three or four each day until Monday when we spoke to the doctor to make appointments for treatment. The doctor ordered Tegretol®, as well as an MRI and an EEG. For the next six months, life continued as normal on her medicine. She continued to learn and grow, and we didn't see any more seizures. We moved our family to Washington State and followed up with a local neurologist to monitor Sissy.

Just after Sissy's second birthday, the seizures came back strong. There were three different kinds that occurred daily at anytime day or night. We tried Klonopin®, Depekene®, Meberal®, and a few other medications. Sometimes the seizures actually got worse with the medicines; other times, the side effects were so bad that it didn't matter what the meds did for the seizures.

Sissy then began to display a wide array of autistic behaviors that became normal to us. Before the seizures, Sissy would sing songs like “Baa-Baa Black Sheep,” “Jesus Loves Me” and “Row, Row, Row Your Boat.” Before the seizures she looked at people, played with her dolls and played “kitchen” with the neighbor kids in the back yard.

After age two and the onset of the seizures, everything changed. She could only remember “Twinkle, Twinkle Little Star” to sing. She flipped pages in books front to back, back to front, over and over again. She became very upset and threw uncontrollable tantrums if you took a book before she was to the end of one side. She carried a deck of cards with her and stacked them one at a time perfectly aligned in the same direction back and forth, over and over. She couldn’t stand to be in a room where there was more than one source of noise such as a TV and a conversation. She would gather her things and go to a quiet corner or room and continue her pattern playing.

During this time, she began having up to 10 grand-mal seizures and countless absence seizures every day. We started to see Dr. Glass in Seattle before Sissy turned three. It was there that I also met the most compassionate nurse, Joyce, who always gave us hope and the moral support we needed over the phone when we would call there tired and frustrated. Dr. Glass and his office staff were a great help and had a lot of experience with TSC.

After several tests, it was determined that Sissy’s tubers on her brain were inoperable. They were spread out and the tests couldn’t tell which tuber or tubers were causing all the problems. Therefore, the medicine struggle continued until one day in June 1990 when she had a day with no seizures at all. We had found the right combination of medicines...for awhile anyway.

I don’t blame TSC alone for the separation and divorce from Sissy’s daddy. But I know that all people handle stress differently and that has a big effect on relationships.

In 1991, I (mommy) moved with Sissy, Monica, and their new half-brother, Ben, back to Arizona. We got reacquainted with Dr. Frost, and Sissy started preschool in my hometown of Gilbert. She didn’t speak well or run correctly so she started therapies, and we continued to try to control the seizures. Sissy had a few wipeouts from seizures where she got facial bruises, rug burns, sand scrapes—usually on her face. She never took a bath or played outside without adult supervision, because we never got a warning before a seizure. She was a happy little girl and the other kids seemed to know she was different but were nice to her and allowed her to play alongside them if she chose to. I remarried and we moved back to Washington in 1993, but that didn’t last long either. Just after our second anniversary I was divorced again but we didn’t move again.

School became harder for Sissy as she got older. School systems weren’t as accommodating as she needed. There always seemed to be a struggle when asking, then demanding, special services from public school. Sissy’s siblings had friends who were good to her and even went out of their way to help make sure she was included in their games. The friends even changed the rules to “UNO” so Sissy could understand it. Sissy liked to help with chores such as making her bed and feeding the cats. We had an overweight cat because Sissy knew the cat would come out of hiding when it heard food in the bowl and then she could hold it! Pretty clever for a nine year old that wasn’t able to read or count all the way to 20.

Sissy learned to tie her shoes at about age 10. And when she was 13, she learned to ride a bike without training wheels! She isn't good at the brakes but the side of the house or car seems to work fine for her. I remarried again when she was 10, and Sissy really responded well to my new husband. She enjoyed his way of teaching her new words or learning new little things about how to do things.

Anxiety and angry behaviors became an issue as she approached puberty. She was unable to focus on more than one adult in the room and would always choose one adult to "pick on." This is a tough situation for anybody after working all day. It became hard to tell if this was simple defiance, a result of seizures, meds, or maybe she had a headache or other abstract feelings. Possibly it was the inability to transition into the situation in the room changing in the evening. Either way, evenings became harder for everyone instead of a family time to relax. My husband and I ran into several roadblocks that we just couldn't fix regardless of our efforts. In fall 2002, it was just me and the kids again and the divorce was final early 2003. This was hardest on Sissy, I believe, but it had to happen.

In July 2003, Sissy had a vagal nerve stimulator implanted. What a powerful feeling to stop a seizure with the swipe of a magnet! But, soon it didn't seem to help. This was our last step before brain surgery, if that was even possible. Her behavior became so unpredictable and out of control that we used injectable Ativan® to control severe behaviors.

There was one day where she even kicked me in the head and made me dizzy for a moment. She had been squeezing the kitty too tight and I had saved it. When she tried to leave to tell the neighbor how mean I was we had to lock the "Sissy locks." These were special locks I had to install for her safety to keep her in the home when she got angry. If she left, she would walk away even into the road or into a stranger's house not knowing it was dangerous. These locks were only used during bad situations and had to be unlocked with one of three sets of keys kept in our home so at least Sissy would be safe as she calmed down.

We also began a journey after December 2002 to save her kidneys. She had developed growths there also and her kidneys were actually shrinking as they were having less good tissue to use. We added more meds and a new way of eating to try to prolong any thoughts of dialysis. So far it's working.

Later in 2003, Sissy began sleeping more and more and losing interest in eating or drinking. Mealtimes became a chore. It was May 2004 when she was 16, 5 foot 3, and down to 93 pounds. The doctor put in a gastric tube, which allows me to provide needed water and supplemental nutrition when she won't eat. When she feels good she eats, and I can still control the hydration and add calories and nutrition without a battle.

Sissy turned 18 in 2005 and we are done with public school. On good days, she likes to learn at her own pace. I still work with her and hope some day that all the letter sounds she knows will just make sense as words and maybe she will read a little book, any book some day.

As Sissy turned 18 I had to obtain legal guardianship through the courts. This was just prior to a big decision. Because her seizures had become more frequent and debilitating, we were facing brain surgery. Dr. Ojemann at the University of Washington was our neurosurgeon and he used a grid to locate the active tuber. A week later he removed it. This was

the most trying, scary, and helpless time of my life. My boyfriend, Lee, my kids, Sissy's daddy, and some members of our TSC support group were by my side through this ordeal and it was a very good thing for me to have that support.

Sissy made it through the surgery quite well, and there were great changes in the weeks after the surgery. Her vocabulary increased, and her understanding of implied situations emerged. She even plays little jokes on people now! She's really very funny sometimes, and she smiles again. I hadn't realized how long it had been since she had laughed.

Sissy went to the mall with her sister that Christmas—a trip to the mall, one on one, just to shop. It seems so simple but with all the noise and chaos there, it was something they had never done. They even agreed on some cute Marines there and Sissy announced it to them as they walked by! There were still seizures but it was getting better. Sissy took horseback riding lessons and later spent time learning the basics of swimming.

It's barely into 2007 as I'm writing this and the last six months have been rough with more seizures, behavior issues and weight loss. She is down from 118 to 103 pounds because constipation mixed with throwing up often. We've been through tests and the bottom line, the culprit, appears to be the vagal nerve stimulator (VNS).

The device was off for about six weeks last fall prior to being replaced when the battery died, which is when the gastric issues began. Either the nerves around the intestines aren't working without the charges from the VNS or the VNS has fried the nerves leading to that area. You see, the vagal nerves are largely responsible for peristalsis and now she has none. This is a daily monitoring chore and requires more meds and close monitoring of everything going in and coming out. I am currently working my last week at my nursing job and using the Family Medical Leave Act to try to resolve this as well as the tonic-clonic seizures that now number three a day. We plan to resolve this soon.

TSC is hard on the whole family. It's hard on finances when one takes short shifts or extended time off to care for a family member. It's stressful on the parents, step parents, and certainly the siblings. It's sad to watch my daughter struggle and to see her feelings hurt as her siblings grow up and get busy with their own lives. It's sad that she will never be a wife or a mom.

TSC is exhausting to all of us. I'm a tough mom, but I'm tired of TSC. Oh, I won't quit fighting for my daughter and for others with this disease...but I'm tired.

Mom's Turn

By Susan Taylor

Our 17-year-old daughter, Chandlee, was diagnosed with tuberous sclerosis complex (TSC) in 1990 at nine months of age. As was the case with so many TSC diagnoses at that time, it should not have taken nine months to identify. Chandlee, although a beautiful and healthy baby born full-term, had seizures from birth and classic white spots all over her body. She was even born with a beautiful tuft of white hair, which I thought was so special.



Although we expressed concern to our doctor about the “spasms” with her neck and right arm as well as the weakness on her right side that interfered with crawling, she was declared perfectly healthy even after having a febrile seizure at eight months of age. It was only because my mother, sister and brother-in-law were in the health profession that we referred ourselves to a pediatric neurologist at Johns Hopkins Hospital for a second opinion. On my family’s advice, we videotaped Chandlee’s “spasms” fully expecting the neurologist to concur that there was nothing wrong with her.

On Halloween, we went to the appointment with our daughter and the videotape in hand. Immediately her “spasms” were diagnosed as seizures, possibly infantile spasms, and the white spots that glowed under the Wood’s Lamp were described as classic TSC skin manifestations. My husband and I were also evaluated with the Wood’s Lamp but had no outward signs of the disorder.

Chandlee was put on seizure medication immediately and additional tests were ordered—EEG, MRI and CT scans—to confirm her diagnosis. By her first birthday the test results showed she had more than a dozen tubers in her brain, abnormal EEG confirming seizure activity (but not necessarily infantile spasms) and a definitive diagnosis of TSC. We were devastated not by the fact that she had a rare disorder that we may have passed on, but because we were told she would likely begin regressing and her overall prognosis was very poor. We were not encouraged to hope for the best, but were rather faced with the worst-case scenario.

Fortunately, we had a very strong and extended network of familial support from both sides. Various family members across the country began researching TSC on our behalf while we continued to videotape and document detailed accounts of seizure activity, blood levels and developmental progress. We were also fortunate that Chandlee’s seizure medication, a combination of Tegretol® and Klonopin®, worked almost instantly. Her seizures abated, she gained the use of her right arm and hand, and she continued to develop according to the normal milestones with a little speech therapy support.

Although Chandlee continued to make progress, we did have many emergencies throughout her toddler years with status epilepticus, which either occurred with a high fever or when we tried to wean her from the Klonopin. Eventually, she outgrew the ear infections that usually caused the fevers; we kept her on the Klonopin until she outgrew the dosage and it was clear that it couldn't be providing any therapeutic advantage. We learned to call 911 immediately when needed and to provide the emergency personnel with a protocol about how to treat her.

From the beginning, we became very active with the National Tuberous Sclerosis Association (aka Tuberous Sclerosis Alliance) so that we could learn as much about the disorder as possible. We wanted to help those with greater challenges than Chandlee and fund research into the disorder in hopes of overcoming future problems that Chandlee might encounter with TSC. We continued with routine blood work, MRIs, EEGs and sonograms, and she continued speech therapy intermittently.

Each school year I briefed all of Chandlee's teachers about the disorder and discussed accommodations she needed. We helped raise money through garage sales, walk-a-thons, raffles and other fundraising events. I assisted with the local TSC clinic, attended conferences and medical symposia, and volunteered in the local parent network.

Above all, though, I wanted Chandlee to understand all the facts about TSC and not be "ashamed" that she had spontaneously acquired this rare genetic disorder that caused seizures and other nuisance symptoms like facial angiofibroma. I wanted her to be confident and comfortable in dealing with any problems she might encounter from the various manifestations of TSC. I wanted her to be able to advocate for herself and explain her disorder to the curious and the ignorant. I felt a great need to educate the public on disability awareness and to change people's attitudes toward all individuals who were "different."

When Chandlee was three-and-a-half years old, our son Peter was born. Ironically, Peter was born with multiple disabilities from brain damage caused by a stroke in utero. His challenges are great. He is non-verbal but speaks proficiently with a high-tech communication device, is now ambulatory but has very poor balance, has fine motor difficulties, is profoundly deaf in one ear, has some vision impairment, has sensory integration dysfunction, exhibits ritualistic behaviors, and is cognitively delayed.

Chandlee is a devoted big sister to Peter and advocates for herself and Peter. She has learned through her struggle with TSC how to fight for all individuals with disabilities. Each year as researchers learn more about TSC and she learns about more challenges she might face, she is a stronger advocate for herself, her brother, and all those individuals with "differences." We couldn't be more proud of her!

Glossary

AD/HD (attention-deficit/hyperactivity disorder) is generally considered to be a developmental disorder, largely neurological in nature. The disorder is characterized by a persistent pattern of inattention and/or hyperactivity-impulsivity. Science recognizes three subtypes of AD/HD (inattentive, hyperactive-impulsive, and combined).

Autism Spectrum Disorder Autism is a complex brain disorder that inhibits a person's ability to communicate and develop social relationships and is often accompanied by extreme behavioral challenges. Autism Spectrum Disorders are diagnosed in one in 150 children in the United States.

Benign Tumors Non-cancerous growths. Most forms of benign tumors do not metastasize (spread to and grow in a distant focus in normal tissues elsewhere in the body).

Cancer Cancer develops when cells in a part of the body begin to grow out of control. Although there are many kinds of cancer, they all start because of out-of-control growth of abnormal cells.

Cardic Rhabdomyoma A benign tumor composed of muscle tissue that occurs in the heart.

CT (computerized tomography) A technique for creating images of the internal structures of the body. CT scans are formed from computerized imagery of many highly precise X-rays.

Cyst A closed sac containing fluid or semi-solid material, developing abnormally in a body cavity or structure. Cysts can be damaging to surrounding tissue.

Dermatologist A healthcare provider specializing in disorders of the skin.

Developmental Delay Delay in the normal cognitive and/or physical development of a child.

Early Intervention A federally mandated, state administered program that provides interventions for children age 0 – 3 years who have or who are at risk of having developmental delays. The programs usually include various therapies (physical, occupational, speech, etc.).

ECG, or EKG, Electrocardiogram A recording created by an instrument called an electrocardiograph showing a record of the electric activity of the heart. This noninvasive procedure shows if there are abnormal cardiac electric impulses and/or rhythms.

Epilepsy When a person has had two or more seizures that have not been provoked by specific events such as trauma, infection, fever or chemical change, he or she is considered to have epilepsy.

Facial Angiofibroma A benign tumor of the face composed mainly of blood vessels and fibrous tissue. Angiofibromas initially appear as pink or red bumps and can form a butterfly shaped distribution around the nose, cheeks and chin.

Genetic Counselor A counselor who specializes in genetic disorders. Genetic counselors help individuals with genetic diseases and their families make medical and personal decisions based on their genetic information.

Genetic Disorder A disease or condition caused by an absent or defective gene or abnormal chromosome.

Hamartoma A common benign tumor in an organ composed of tissue elements normally found at that site but that are growing in a disorganized mass.

Hypopigmentation Skin abnormality featuring less color, or pigment, than normal. In TSC, hypopigmentation appears in the form of spots, or hypopigmented macules, on any part of the body. These spots are benign and pose no physical threat.

Infantile Spasms A severe type of seizure that typically occurs between the ages of two months and two years, although most children who develop this type of seizure are around 6 months old. It is identified by sudden myoclonic jerks, flexing of the body and neck and stiffening of the limbs. Each of these seizures lasts a very short time, but can occur in long or short clusters. If left untreated, infantile spasms can have a devastating effect on a child's intellectual development.

Lymphangioleiomyomatosis or LAM This is a lung disease that is caused by mutations in the TSC genes that can occur in individuals with TSC, primarily women, or in sporadic cases. Cystic lung destruction leads to loss of lung function in LAM.

Malignant Tumor A cancerous tumor.

Metastasis Sometimes abbreviated Mets, is the spread of cancer from its primary site to other places in the body (e.g., brain, liver).

MRI, Magnetic Resonance Imaging A non-invasive system producing images of brain tissues by using radio waves and strong magnetic fields. MRI can detect tumors, tubers and other soft tissue abnormalities.

Neurologist A healthcare provider who specializes in the function and disorders of the nervous system.

Neurosurgery Any surgery that involves the brain, the nerves or the spinal column. Neurosurgery of the brain may be performed in an attempt to control seizures, to remove a brain tumor or to alleviate the pressure from hydrocephalus.

Ophthalmologist A healthcare provider who specializes in the functions and disorders of the eyes.

Polycystic Kidney Disease (PKD) Polycystic means multiple cysts. In effect, PKD denotes multiple cysts on each kidney. These cysts grow and multiply over time, also causing the mass of the kidney to increase. Ultimately, the diseased kidney shuts down causing end-stage renal disease for which dialysis and transplantation are the only forms of treatment. PKD comes in two forms. Autosomal Dominant Polycystic Kidney Disease (ADPKD) is the most common, affecting 1-in-400 to 1-in-500 adults. Autosomal Recessive Polycystic Kidney Disease (ARPKD) is far less common, affecting 1-in-10,000 at a far younger age, including newborns, infants and children.

Seizure In normal brain function, tiny electrical charges pass from nerve cells in the brain to the rest of the body. A seizure occurs when the normal pattern is interrupted by sudden and unusually intense bursts of electrical energy that may cause strange sensations, emotions, behaviors or convulsions, muscle spasms and loss of consciousness. These unusual bursts are called seizures.

Shagreen Patch Abnormal patches of skin resembling an orange peel, usually found on the lower back or the back of the neck. Shagreen patches may be present on other parts of the body as well.

Subependymal Giant Cell Tumor (SGCT) A benign tumor found in the brain of individuals with TSC. SGCTs typically grow near or in the ventricles and can cause hydrocephalus (increased pressure in the brain) if they block the flow of cerebrospinal fluid (CSF).

Subependymal Nodule A non-cancerous nodule (collection of cells) located along the edge of the brain's ventricles. Subependymal nodules can grow into subependymal giant cell tumors, and some subependymal nodules become calcified (filled with a calcium deposit).

Tuber An area of the brain that contains a disorganized collection of abnormal cells; usually found in the outer layers of the brain called the cortex, but can be found in deeper areas of the brain.

Tumor Tumor is primarily used to denote abnormal growth of tissue. This growth can be either malignant or benign.

Ungual Fibromas Benign fibrous tumors found in the areas around the fingernails and toenails.

Wood's Lamp An ultraviolet light used to detect hypopigmented macules in TSC, and used to diagnose other skin and scalp diseases.



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