

June 15, 2012

Tommy was first diagnosed in 1995 with a TIA, which sent him to the hospital with arm and leg numbness and extreme fatigue. He was 42 years old. The doctors performed an MRI and said he had MS (Multiple Sclerosis).

A few years previously, my mother in law had passed away from a stroke at the age of 57, which was the final in a series of strokes, and TIA's that were diagnosed by the Mayo Clinic in Scottsdale Arizona as "Binswangers Disease."

At this time, no one had heard of CADASIL, including Tommy's neurologist and me. I had a very good friend who had MS at the time and I just got an uneasy feeling that this was not MS that Tommy had because it was somehow different although some aspects were similar at the time. Therefore, I began doing research on the internet and at the Library of Medicine in Bethesda, MD near the National Institutes of Health, which I am fortunate to live nearby. I began corresponding with a PH.D Hematologist (blood doctor) who was doing research on rare brain diseases and was currently in France. It was quite by accident that I found her name.

She told me about CADASIL and that it was possible that Tommy had this disease and had inherited it from his mother. Tommy's mother had a brain biopsy done before she passed away from a stroke and Palsy on her face and the Mayo Clinic still had the brain tissue. The Hematologist asked me if I was willing to send the brain biopsy and a sample of Tommy's blood to her so she could use both in her research and she could then study the DNA and the gene patterns to see if Tommy had CADASIL which at this time was only diagnosed by a skin biopsy and I could not find anyone to perform this test except for in Europe and this was out of the question for Tommy and I to fly to Europe as we did not have the funds to do this and we could not find anyone in the US to give him a skin biopsy for this purpose. The Hematologist also informed Tommy that if he had siblings they could send their blood along as well and this would be very helpful in her research but she would not reveal the results if they did not want to know if they carried this disease. Tommy has two brothers, one consented to send a sample of his blood, and the other did not want to know and did not consent to sending his blood.

The results came back as having the CADASIL disease by the DNA patterns and the NOTCH locations etc. that is typical of CADASIL. She also discovered another genetic deficiency, which would make it impossible for Tommy to ever be put under anesthesia for surgery, as he would probably not wake up. I cannot remember the name of this genetic issue at this time. Tommy had a severe nasal infection and needed surgery on his nasal passages, which was due to happen in a few months. We cancelled the surgery and now just put up with this nasal problem, which causes alot of snoring and sleep apnea sometimes as we do not want to take the risks with surgery. I later read articles about CADASIL and sinus problems being connected somehow in some cases. Tommy's brother (that we sent the blood) did not have CADASIL but his mother definitely did. I called the Mayo clinic and informed them in case they needed to know for research purposes as well.

I found Billie by miracle before her website and before there were many CADASIL people that connected online. I remember corresponding with Billie by email about Steve and it was a great help to me to know someone else was out there. I also spoke with others and particularly remember women in Canada whose husband was in final stages of CADASIL and we would talk by telephone and offer help to each other. Around the year 2000, Tommy was not able to work anymore as he worked as a satellite technician for cable television and was the person responsible for calibrating the satellite dishes and other equipment. His memory was giving him many problems and he was having some speech problems then too. At the risk of having Tommy fired as I did not think the company wanted to deal with a "disabled" employee, I told his supervisor to call me if ever he was walking funny or not making sense when he was talking and I warned

him that Tommy had a history of strokes in his family. One day, Tommy's boss called me and said Tommy was walking, as he was drunk and just did not seem right. I went right away, picked Tommy up, and took him to the hospital. They told me he had a stroke and was having numbness on one side of his body. Tommy luckily was very stubborn and refused treatment then as he was starting to feel better already and get back feeling on his numb side. I look back on this now and see this as a good thing that we did not allow them to administer the typical "stroke" treatments of the time that could have caused Tommy bleeding in his brain.

Throughout the next few years, Tommy would have TIA's, and some strokes and the symptoms he experienced was "migraine without pain." We found this out from a trip to the Mayo Clinic in Scottsdale, AZ where we discovered a doctor who did know about CADASIL (this was in 2005) and Tommy had an Aunt and Uncle who lived in nearby Phoenix that allowed us to stay with them while Tommy was examined by this doctor. He said Tommy was having migraines when he would experience nausea, vomiting, and sleeping for sometimes up to 36 hours at a time, but had NO headache pains. This was a huge surprise to us because we had read that CADASIL causes migraines but Tommy never had one with the pain. However, he had all the other symptoms! Tommy was also a smoker at this time and Mayo Clinic doctor told Tommy he must stop smoking as this causes CADASIL to worsen much quicker. Tommy did not stop smoking at this time.

In 2009, Tommy had a very large stroke and could not move his arm or leg on the right side of his body. This has stuck with him to this day and he is now confined to a bed most of the time with muscle aches and pains (part of the muscular dystrophy effects of this disease) and the inability to use the right side of his body. Tommy is also incontinent and needs a lot of care such as assistance with eating, hand coordination, emotional ups and downs, etc. I am also fortunate enough to be healthy and care for him myself and work full time right now. I hope and pray every day that I will be able to keep this up for Tommy's lifetime as I cannot put Tommy in a nursing home. I think they are awful places although I know for some it is the only way to care for an extremely ill and/or frail individual.

I am telling my experience because when Tommy first was diagnosed with CADASIL back in the late 90's, Tommy and I sat down and examined our lives and our future. We decided to take some trips to places we had always wanted to go but were waiting for retirement to do. We realized then that we would not reach retirement together and that it was going to get harder and harder to enjoy our lives as they had been in the past 20 years we were married. We bought some items that Tommy really wanted to have like a large screen T.V. and a riding lawnmower. We did not have the money at the time, but had the ability to borrow money to do these things. We also were not able to have children together (although I had always been very upset about this) so we did not need to worry about passing this on to others in our immediate family or caring for young children.

We did not deny ourselves things within reason and enjoyed life to the fullest. We are very thankful that we did these things while we could enjoy them together. We know this is not possible for some who receive this dreadful diagnosis of CADASIL, as it is too far advanced, but we hope others will see the warning signs, have strength to get a diagnosis, so you can also enjoy life to the fullest while you can. As another note, Tommy has seizures and TIA's all the time, but has told me not to take him to the hospital and not to treat him for any other medical conditions. This is his choice and I respect that. It is hard for me to accept this, but I understand it. He does not want to be an "experiment" and/or have test after test, which leads to nowhere right now. He won't have any more MRI's as there is no point to these anymore as they just tell him he is getting worse.

Sometimes we have to do what a loved one wishes and knowing that we are upholding their "choice" is true love! Thank you for listening to our story.