Mike and I were married on September 25, 1993. The very next day, Mike's dad suffered his first stroke at the age of 64 after years of good health. He recovered reasonably well and went on to enjoy his first few years of retired life. A few years later, Mike's parents moved to the West Coast. The move was very difficult for his dad who apparently suffered numerous Tia's during the transition. Physically, he slowed tremendously, shuffled when he walked and slurred his words when he spoke. . Frequent visits to the doctor found that he had continued to suffer mini-strokes but nobody could figure out why. He has slowly declined over the years but is still living independently and doing pretty well at age 76.

Mike has always been an avid athlete, particularly a hockey player (yes, a true Canadian), sometimes playing three times per week. He has never smoked, his blood pressure and cholesterol are normal; he eats well and is very active. We had 2 young boys with a third on the way when four years ago, he was at work one night and started to feel numbness down his left leg, foot, arm and left side of his face. He came home right away and we went to the hospital. After a few hours his left arm was totally recovered but he continued to experience numbness and tingling in his left foot and left side of his face. He had a CT scan which showed some abnormalities which was followed by an MRI a few weeks later. When we met with the neurologist to discuss the results of the MRI, we were told that Mike had Multiple Sclerosis. We were shocked, upset and scared, especially with our third child expected in a couple of months. He immediately started on Avonex, a weekly injection of Beta Interferon, one of the treatments available for relapsing-remitting MS. Learning to give himself the shots was a challenge. Each week I would know that he had succeeded when he would reappear after locking himself in a room for a good 30 minutes, only to come out as green as grass.

After three and a half years of weekly injections, he had no further "relapses" of his MS until last Christmas (2005). On the night of Christmas Eve, he first noticed feeling as though he was slurring his words when speaking which progressed to immense difficulty with speaking within a few hours. Assuming that this was likely a "relapse", we didn't panic too much and decided to get a good night's sleep and hope that he would be better in the morning. And, given that it was 11:00pm on Christmas Eve, with three young children in the house eagerly awaiting the arrival of Santa, we took a very relaxed approach to his symptoms. By 8:00am the next morning, he couldn't speak at all. He could cough but that was really the only sound he could produce. Right away, we went to the local ER. As the day progressed he noticed that his right side was also affected by weakness and poor co-ordination. By that night, he couldn't walk without assistance. The ER doctor also assumed that this was just a relapse of his MS and started intravenous steroids to aggressively treat the relapse. He had 5 days of IV prednisone and then another 3 weeks of oral treatment. One nice thing was that all of the treatments were done at home so Mike didn't have to be hospitalized through the holidays.

After the first week, he started to improve physically. With the amazing help of OT and PT, his walking recovered almost 100%. We were surprised with how well he recovered, given how bad things had seemed only a few weeks before. However, his speech did not improve very much. By the end of January, he was able to say some words but it was very difficult to understand what he was trying to say. He tried to write, but of course, he was right handed

and this was very challenging for him. Eventually he was given a "communication aid" – basically a palm pilot that will speak what you type into it. He was never the best typist and he found it very difficult to use. Also, he felt strongly that he needed the practice verbally and when he relied on the device, he didn't get that practice. He continued with speech therapy at least once or twice per week and to this day he still practices regularly and continues to hope that his speech will improve. I notice that he has good days and not-so-good days now but overall, his speech is a major disability. We are still hopeful that it will recover! The other day, our eight year old asked if we had any videotape of dad speaking because "he can't remember what he used to sound like".

In February, Mike's neurologist continued to question his very unusual presentation of multiple sclerosis. We went for a second opinion to a downtown MS clinic and before we went, the neurologist asked me to ask them about the likelihood of CADASIL. This was the first time anyone had mentioned the disease to us. When we asked the MS specialist about CADASIL, his response was "you're talking one in 35 versus 1 in 300,000 – not likely". After reviewing Mike's MRI with a neuroradiologist, he returned to inform us that together they felt there was less than a 5% chance that this was anything but an unusual case of MS.

Still not satisfied, Mike's doctor ordered a skin biopsy just to rule out CADASIL. Around that time we did an extensive search on the internet to learn more about CADASIL. One of the first things we saw was the occurrence of migraine. To this day, Mike denies ever experiencing a headache, never mind a migraine! We had convinced ourselves that CADASIL just didn't fit Mike's case – NO WAY.

Until we received a shocking phone call in June of this year. Mike's skin biopsy had shown granular osmiophilic material indicative of CADASIL as the correct diagnosis. Since then, we've looked further into Mike's family medical history. It turns out that Mike's grandmother (father's mother) suffered from "hardening of the arteries" and died of Alzheimer-like symptoms in her mid-60's. She was one of 4 siblings, 3 of whom appear to have also had the disease. Four other descendents of their family have "M.S.".

Mike has just had the genetic blood work done to confirm the exact mutation in his family. After we have these results, all of the other interested family members will also be tested. Mike is the oldest of 5 siblings so chances are that he's not the only one with the disease.

We were devastated to learn about CADASIL, particularly the autosomal dominant part with three young kids, ages 11, 8 and 3. We've decided not to test the kids and let them make up their own minds about testing when they are adults. It is our hope that by that time, there will be at the very least a treatment for CADASIL and hope for future generations. We look forward to learning as much as we can and doing whatever we can to help raise awareness of CADASIL and helping out in any way possible.

Lorraine Anderson
September 9, 2006