HOW MY HUSBAND STEVE IS SURVIVING HIS TERMINAL ILLNESS CALLED CADASIL

This is my story about CADASIL, how I found out my husband has CADASIL and why I established the web pages.

This is how my husband described the events of the morning he woke up: "waking immediately in the early morning of November, 1994, I had the most excruciating, mind splitting headache, and I sprung out of bed. Although in the worst pain of my life of 38 years and in total darkness, I slid out of bed and crawled to the bathroom. Finding the commode, I vomited violently. Still in heavy pain and my eyes not able to focus, I repeated the vomiting process as the chainsaw was cutting into my head and the hot molten lead poured down. Some relief was near as I pulled myself to my feet. I used every bit of energy I had left to rise up to a standing position. I fumbled to switch on the bathroom light. Instantly, I was in violent pain. I vomited into the sink and tried to crawl back into bed."

My husband had always been in great health and has had perfect attendance at work until 1994. I do remember my husband's words, "Just let me die, as the pain is so bad". What could I do to help him? What does he have? Tests were performed, with no answers. They even said that it might be in his head and sent him to a pain management clinic. In 1995 he had another major attack, which placed him in the hospital. This time the CAT scan came back abnormal. An MRI was then performed with the results of white matter and lesions. Immediately, they referred us
to a Neurologist. The words from him were very supportive at first, but not promising. He said, "I cannot believe this, you look too good to have the MRI come out this way. We will definitely start to rule out Leukodystrophy. MS has migraines, maybe dementia or it could be in his head. We will find out what this is, even if we have to research more or even if we have to use Lorenzo's oil. We came out of the doctor's office, stunned when the Neurologist's words did not give us any hope.

February 1997 Steve was functioning the best he could, never complaining during test after test, finding no answers or no relief from migraines shots, jab, CT and MRI's
Spinal taps, etc. and nothing helped. I remembered the words of the Neurologist about"Lorenzo's Oil", so my husband and I watched the movie. I was no computer wiz but within a week I bought a computer and started doing research. To this date, I have replaced the computer three times and Neurologists three times. Prior to the Internet being popular and with no training, I looked up excessive white matter disorders and anything I could from the results of the MRI and CT scans. I got copies of my husband's files, converted them to MS Word, and sent over 200 E-mails out with my husbands' history and asked for help. Our first neurologist was so overwhelmed and amazed with the support and response we received. I had great response from doctors, out there on the World Wide Web. I received a call at work from Dr Raphael Schiffman, National Institute of Neurological Disorders and Stroke, National Institutes of Health, Bethesda, Maryland, stating that my husband might have CADASIL, however, they did not know too much about the disease.

What is this CADASIL and what does it mean? CADASIL is an acronym for Cerebral Autosomnal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy. CADASIL is a diffuse disease of small arteries predominating in the brain. It starts during mid adulthood and is characterized by recurrent alchemic events (transient or permanent), attacks of migraine with aura, severe mood disorders, and subcortical dementia and, at MRI, a white spread Leukoencephalopathy, TERMINAL ILLNESS AND AN INHERITED DISEASE. CADASIL does not have a treatment or a cure. I cannot describe how I felt. I cried. I was mad. How can this be? My daughters also have a 50% chance of having CADASIL. There were about three items on the Internet concerning CADASIL. The information related so much to my husband's symptoms. I was hoping that it was not CADASIL and maybe it was something else.

Dr. Hugo Moser, (known for Lorenzo's Oil) of Johns Hopkins University and Kennedy Krieger Institute had answered my e-mail and assisted us in having blood drawn and sent to him to rule out a Leukodystrophy. I Fed EX'd the MRI, CT Scans, and all other documents to the Kennedy Krieger Institute to aid in his research. An email was sent to me from Boston letter me know about research and gave me the researcher name Professor Marie-Germaine Bousser. The Professor called me and requested copies of my husband's MRI and medical records that included my husband's family history. Through the Internet and by fax machine, I located medical records from my father-in-law's doctors going back to the 1960's. These were forwarded to the Professor in France. The Professor informed me that my husband needed a rare skin biopsy, and she sent the instructions on how to have an electron microscopy skin biopsy. It took over 2 months to locate the medical supplies, a dermatologist to perform this procedure, and a radiologist to read the results. I did not stop calling to locate professionals who were willing to perform this test with the instructions I had obtained from France.

Concurrently, Dr Moser had met with one of his colleagues from the Netherlands, having all the original documents and films from my husband's case and was confirming my husband had CADASIL. At the Age 40½ years old in July of 1997 (2 days before the 2nd anniversary of my father-in-law's death), the skin biopsy test

confirmed CADASIL. My Husband and his father, who died in 1985 from a stroke, were both confirmed with having CADASIL. CADASIL is passed from parent to child through a mutated gene. Each offspring of a CADASIL parent has a 50% chance of inheriting the disease. If a person does not inherit the CADASIL gene, he or she will not develop the disease and cannot pass it to subsequent generations. A person who inherits the CADASIL gene will sooner or later develop the disease. You can only treat the symptoms, as there is no treatment or cure.

How are we going to cope with this? My husband, at the age of 40 years old, has a terminal illness, and my daughters might have it. Why could it not be cancer or something with a least a treatment? Steve's mother had to take care of his father for 5 years with no assistance. The doctors could not say what he had. First they said cancer. Then they said that he may have dementia at 48 years old. Finally, they told her to take him home and make the best of it. He died at 52 years old. How can we tell his mother that her son, my husband, has the same disease that her husband died of, and that Steve's sister and three other brothers, might also have the defective gene?

What can I do to help him? It hurts to see my husband in pain. I contact the doctors or to cope I do more research on the Internet to find more information on CADASIL. I have doctors and others e-mailing me wanting to know about CADASIL. This is how I cope- when I come home from work, I get on the internet and hope that I can find someone who can help us learn more about this disease. Our children ask if daddy is going to get better. How do you answer? I am scared that he is going to die young like his father. Our children know that their daddy is always going to be sick for the rest of his life, and we have to learn to cope with it. I love my family and am scared of losing my husband and children. I want to protect everyone.

When Steve was in the hospital in 1999, our youngest daughter, Noelle, finally talked to me about her Daddy. She asked me if Daddy was going to die. I had to explain the disease and how her granddad had died of CADASIL. She asked me if she had it. How I wanted to lie and say no. I explained that the symptoms usually do not show up until you are nearly 40 years old and she has a 50% chance of having it. Then we talked about how Daddy has donated blood to Dr. David Lynch in Penn University to create the first blood test for CADASIL. Hopefully they will have a test when our daughters get older. She asked if there was going to be a cure. I explained that I hope to live until they do. But how about daddy and a cure? It is so hard to talk to her about this. I explained that if they don't have a cure for daddy, your daddy is going to donate his brain to science so when you get older you can say your daddy helped you.

Oh, what a wonderful thing to give to his children. Other children may get money, a car, etc. But what a legacy they will have. We can get pictures of Mars, have medicine to treat cancer, even to help with AIDS, but there is nothing for my husband's rare condition. They can only attempt to help with some of the symptoms. We went to a big medical center 4 hours away in Houston. With all the medical buildings and science centers, no one knew about CADASIL except one doctor. After seeing a director about strokes, he turned and told me that I knew more about CADASIL then he does! How can this be? We live not knowing when my husband's next TIA (small stroke) will be or whether will he survive it. He has had at least one to two TIA's per year that we know of. He did have a stroke in 2004, which left him weak on one side.

Inserts from Journal

My husband, my daughters' and I feel we are living in a nightmare and would lie to express our appreciation for reading this story. Our hope is to link to others who might be in a similar situation to or. We take people for granted and until this happened I knew I loved my husband but did not know how much until now! Thank you in advance for your assistance with this matter. Thank you in advance for your assistance with this matter. You from a scared wife and mother, Billie

August 1997 – It has been a month since Steve was diagnosed with CADASIL. My husband worries about our two daughters and me but when he is so much pain he feels bad for us. He thinks about his father, how he died so young and how things make sense now on how his father suffered. What can I do to help him? I feel inadequate and it hurts to see my husband in pain. I contact the doctors or try to research, this is how I cope. I give him a heating pad to place on his head. I worry. There is a big chance of my children having this disease. Will they be like this at my husband's age? Our children ask, is daddy going to get better. How do you answer, as I am scared that he is going to die young like his father? Our children know that their daddy is always going to be sick the rest of his life and we have to learn to cope with them.

In 1997 at my daughter's elementary school, I volunteered to create a website. Not knowing at the time, but as I reflect back now, I was establishing and validating my own skill sets and developing the courage to go forth to put CADASIL out for the entire world to see. Everyone who helped and listened, I sent a personal letter and a

link to the website with a big heartfelt thank you. I am not in this for any profit or personal gain. The website was solely set up for information reasons, an on-line support group and network for all sufferers and "Together We Have Hope" website grew. The demand for information had increased; I compelled to create a newsletter. The first newsletter for "CADASIL Together We Have Hope" was mailed out on February 14, 1998. . The response was a fantastic. This went from helping others to an online support group. No words could express the amount of thanks to everyone who had contributed to the website and newsletters. The website was name CADASIL Together We Have Hope.

1998 – Received this information from Paris: The most up to date research on CADASIL is being done in Paris France at Hospital Saint:Antoine, by Professor Marie Germaine Bousser and Dr. E Tournier:Lasserve Unite De Recherces De L'Inserm 25 and their staff. They were the first to diagnose CADASIL. As of February 17, a number of research approaches are currently used in our lab to understand the mechanisms. Underlying the occurrence of vascular lesions in CADASIL, with the final aim to be able to prevent them among these various approaches we are currently making a transgenic mouse expressing CADASIL mutations with the hope that it will be a model for the disease and that we could use it to check drugs. When any of these research approaches will provide useful information for patients care, we will let you know immediately. Although I am quite confident that our group or one of the other groups working in this field will one

November 1998: this is the usually time of the year Steve gets very sick with bad migraines and nausea. The good news is the prevention medicines my husband is on seems to be working. He has a low-grade headache nearly all the time but no major attacks within the last two months September and October. I am even scared to report this as before September, Steve was averaging an attack per month. We call them attacks as we do not know what else to call them.

February 1999 : This web site is now listed on most well know search engines as CADASIL it has had wonderful results. I never thought this web site would reach all around the world. I wish to thank everyone for his or her contributions, if it was not for everyone's the support would have never happened.

Year 2000 : We registered with NORD – National Organization for Rare Disease and CADASIL have been finally recognized including our website.

Feb 2000 : Sometimes I wish I could wake up from this and everything will be normal. Steve seems so weak sometimes and with his loss of weight his skin feels so different. His skin just seems to hang. Steve has admitted to me he feels he will not see his daughters fully grown and gives in to them too easily. His own neurologists told me he does not know what else he can do for Steve.

Received an e-mail: When I first started reading the messages on this Website I wondered what the future held for CADASIL patients, but now feel sure that research is going ahead full power. Thank you for all your hard work.

September 2000 : A Doctor David Lynch from Pennsylvania University called me the other night. He first apologized to me, as three years ago when I sent out more than 200 e-mails for help, he could not help me and sent a generic response back. He has seen five to six people with this disorder. Dr. Lynch is now requesting my help in developing a test for CADASIL through DNA testing. I posted on the website and newsletter to get patients. Steve's blood was drawn on August 28th and sent. Is this not wonderful a doctor kept my phone number and information and remembered about the search on CADASIL?

October 2000 – Researched on the Internet and contacted any doctor who had e-mail address within a 4:hour drive away that might know about CADASIL. A director of stroke from Houston Texas responded, he had heard about CADASIL and would like to see us. My husband was only 44 years old at that time.

October 2000 : Please e-mail you updates on your progress with CADASIL. Also, if you would like to tell your story (we can leave out your names) please send this to me. It seems to help to tell others. This is how I cope with my husband's illness. When I do the newsletters I sit at the computer and sometimes cry but it does seem to help with the pain and stress.

In the year 2000 under all known search engines for CADASIL there are about 25 documented articles related to CADASIL. I sent an e-mail asking everyone to please let me know if I can publish your name and address and

phone number on the web site. I have never had a crank call since I established the web site and feel that it is a benefit to publish this info. I respect your privacy and only want the best for everyone.

Dr. Colony from the University of New York contacted me to let others know that there lab is beginning to develop molecular DNA techniques needed to diagnose CADASIL.

We can get pictures of Mars, have medicine to treat cancer, even to help with aids but there is nothing for my husband's rare condition. They can only attempt to help with some of the symptoms. When we went to a big medical center 4 hours away in Houston, with all the medical buildings and science center's and there no one knew about CADASIL except one doctor. After seeing this PHD he turned and told me that "I knew more about CADASIL then he does"! How can this be?

I have not stopped searching for answers or hope, especially in Texas. We live in Round Rock about 15 miles from the state capital of Austin. My husband is getting worst and his neurologist does not know what to do for him. If only we could find a doctor who knows at least something about CADASIL!

Steve started with Migraine on August 14th and it lasted for nearly 5 weeks. Can you imagine anyone living with that for over 22 days? In the end through me being the caregiver his was admitted to the community hospital as classified as migraine status. He was given steroids, Morphine and Demerol shots for pain. He was admitted on a Wednesday morning and released on the Saturday. No pain in the head anymore. He was so weak though and looked so pale. Steve has now started to have tremors. His chin, legs, shoulders and especially his hand shake out of control. From the support of this website, I contacted some doctors upstate and they say it likely CADASIL. Steve just turned 44 years old. He finally started back to work part time after 6 weeks of being off. Steve's blood sugar is also not too good at this time. I do wish to thank personally all our friends and coworkers where he works for the wonderful support and understanding. Without this I do not know where we would be.

Received many e-mail similar to this one- First let me thank you for establishing your web site : searching for Information about CADASIL has been difficult. I know two years ago there was very little if anything about this disease on the Internet.

May 2000 : Steve does not complain but he does wake up with a headache each day and really seems to never go away. He does say now he has trouble holding objects for a long time. Sometimes I wish I could wake up from this and everything will be normal. We just had our 20:year anniversary. I love him and do not want to lose him.

To see what type of symptoms or common factors were known. No medical proof but a person who wants things in order was typical of CADASIL including back problems.

Steve has his sugar diabetes under control and in December 2000 we found out he has sticky blood that means his blood clots easy. He is on blood thinners for this, which could interact with CADASIL.

December 2000 : Steve has not been able to go to work since August 14, 2000. After seeing a stroke specialist, he has had a TEE (Transesophagel Echocardiogram) and it came out all clear (Nov 28, 2000). This is where they use a scope down the throat to check out his heart. His heart is good and strong.

March 2001 : Planning is essential I now legally possess Steve's total power of attorney. We mourn each day as we see how CADASIL takes away a little of the person we love each day.

Mar 2001 : I was contacted by a person who asked not to say her name but in British Columbia, Canada the Molecule Diagnostic Lab, Department of Pathology Children and Women's Health Center Address: 4500 Oak, British Columbia v6H321 Canada has a genetic testing for CADASIL. Phone number 604:875:2852.

March 2001 : The Company he works for have been wonderful and understanding, we are very lucky and fortunate. It has been a nightmare and I have not stopped worrying. Steve's gait was off, his migraine hurt so bad he seemed to be hard of hearing. I have noticed something, which I think does not help the progress of CADASIL, stress. Stress seems to bring on an attack or a day later. I have had wonderful support from everyone including the e-mails. Steve suffers everyday with headache migraine and we keep going on. Life is so hard when you have CADASIL, as it is not only a disease that affects the person but it is a family disease and we have to live with it. Someone wrote to me and said that "each day she would hope that her husband would get out of bed and just try to go to work", (this was the same feeling I had) and then everything would be alright. I will press forward.

I worry so much when I am at work. What happens if my youngest daughter comes home from school and finds her daddy dead? I have to try to focus on my work. My sister told me that I have turned a negative thing into a positive thing.

May 2000: I wrote to the news media in hoping to get anyone interested in CADASIL as this is so frustrating that no one knows about it, and that is has never been put in the medical records yet. I want to get the word out about CADASIL as "Together we do indeed have hope".

March 24, 2001 : Steve had his first noticeable TIA. Things started around 6:30 with right eye pain, then numbness on his right side of tongue, into his lips which were light purple, then right side of his face, moving to right side of nose and then all over headache. On his is right hand, three fingers went numb and cold then moved to his other two. The Sensations moved through his hand up his wrist then stopped at his elbow. It was total numbness. Prior to the numbness moving to his lips, he took an aspirin.

August 2001 – was contacted by Paris, France to post the most current information in the newsletter on the ACF:France Cerebral Familial Arteriolopathies Association) Registered office- Hospital Lariboisière – PARIS : MEDICAL DISCUSSION WITH THE FRENCH RESEARCH TEAM DURING ACF'S GENERAL ASSEMBLY ON NOV 17TH 2001 on CADASIL

February 2002 : Steve's had another MRI and they say no change. His balance is off and cannot walk too far without getting so tired. He lives constantly with headaches and gets so weak. I feel sometimes he is like a time bomb ready to go off (and we live each day wondering if he will have a stroke today). His medicines treat only some the symptoms. We are presently starting to file for disability. You see he has not worked since October of last year only two days in January. Disability is very stressful in itself, I try to take away the stress for us, but it's so hard. We try to live each day as normal (ha ha ha)

May 2002 – This is a letter of which my daughter wrote- "My name is Noelle and I am in the eighth grade and I live in Texas. My dad has CADASIL, this illness has affected everyone one in my family in their own different ways. I know this illness has affected you in your own personal life but what I've learned is that all the pain and stress will make you a stronger person for the future".

Noticed that others sites where placing links to my website for support.

June 2002 : I am still looking to go public with the disease here in the USA but no one will listen. This will not stop as I feel I need to go public with CADASIL. We went to Houston Texas for Steve's check:up with the director of stroke. He took blood work for Steve's sticky blood syndrome. Steve has short-term memory loss now. He walks with a cane.

August 2003 : I see Steve so sick, weak and get so exhausted so fast. He does not go with me shopping as the noise and concentration around him is so much. He sleeps half the day and then gets exhausted by just walking around the house. He gets very short tempered and is easily frustrated. His short-term memory is getting worst.

July 2003 : Steve is not doing well. Each week I see a difference in his memory and his gait. It is getting harder to write about how he is doing as it hurts so much to see him looking so old. He stays home most of the time as crowds bring on a migraine or attack.

August 22, 2003 Steve had a TIA on Friday it lasted about 10 minutes. He had numbness started in his tongue then on his right side of his face and then traveled down his arm. He took an Aspirin and it stopped. It left him so weak and exhausted. People ask, How do I cope, how do you work full time?

Saturday, October 18, 2003: Steve had another mini stoke on Saturday morning. He woke up lightheaded, and within one hour told me he felt like he was drunk, which included blurred vision and he was woozy. By the evening, he was having double vision. If he closed one eye, that would stop the double vision. He also felt queasy due to his double vision; we thought it might be a new medicine he was on.
Sunday, October 19, 2003: Steve woke up still having double vision. He made a patch for one eye since he got tired of squinting. By the evening he was exhausted. I sent out a Yahoo Group e-mail to the CADASIL group to see if any one else experienced double vision. I also contacted his neurologist, who was not on duty at the time. The other neurologist told us to go to the emergency room. Steve did not want to go since we previously had been told that there was nothing that could be done for him. I was insistent on going. We were in the ER at 10:00 p.m., and at 1:00 a.m., he was admitted for observation. I carry the CADASIL information in my car, and gave this to the doctors. I also gave them the list of the 13 medicines he is on. When we got to the ER

and we said he had a possible stroke, they asked why we did not call the ambulance. He had a Cat Scan, which showed nothing. All his other neurological signs looked good, but his eyes started to go cross-eyed more. The ER doctor told us he would contact our neurologist in the morning and see what she thinks we should do. FROM NOW ON DO NOT WAIT TO GO TO THE E.R.

Monday, October 20, 2003: When I got to the hospital, Steve had not yet seen a doctor. I went ahead and called his neurologist, and the nurse stated that the doctor does not practice at the hospital we went to. She also stated that she had talked with the ER doctor and told him to monitor Steve's blood levels to do his sticky blood syndrome, but nothing about his eyes. We called back his neurologist, and finally I got to talk with her. She had told the ER doctor that Steve had a mini stroke in the brain stem, which causes the double vision. She also said he needs to patch the eye and every other day swap eyes with the patch for one month. If it does not get better, then they will get glasses for him with a prism to assist him in seeing. We have to wait to see if his eyes will get better by themselves first. Please remember you have to be your own case Manager. We could have still been at the hospital unless I was active and had insisted on talking to his neurologist. It is so hard to hear the words – there is nothing we can do at this time. I am sick of those words. It makes me sick. I am already so upset about the whole deal. Life is so unfair. Seeing Steve lose more each day is so awful. He gets upset so easy and gets so frustrated. When we left the hospital, I even said how lucky it was that only his eyes were affected. It could have been his speech or limbs. He said he would rather have his speech slurred then go through this with his eyes. He lives each day with pain. I keep trying to be brave, but how brave can one person be. It so hard to try to go on and live each day. Tears come so easily and it hurts so much. Steve says things he does not mean. Thank you for reading this, and I hope I can help others like other people have helped me.

August 2003: I see Steve so sick, weak and get exhausted so fast. He does not go with me shopping as the noise and concentration around him is too much. He sleeps half the day and then gets exhausted b just walking around the house. His short-term memory is getting worst.

A movie was just released called the "Sea Inside". This film was the real: life story of Spaniard, Ramon Sampedro, who fought a 30:year campaign in favor of euthanasia and his own right to die and is directed by Alejandro Amenábar. Ramon's girlfriend had CADASIL. Two people had signed the guest book to say they had watched the movie and were wondering what CADASIL was and went to the CADASIL Together We Had Hope Website. Once I came back from the conference, I was searching how many hits I had received on the website for the year 2004. It was unbelievable! There were 200,530 hits on the website.

Feb 2004 – Steve had a MRI – They say: Again identified is much greater than expected white matter signal hyper intensity, both superficial and deep white matter, confluent, anterior and posterior cerebral hemispheres, extending to the level of the convexity superiorly, and inferiority to the level of the occipital and temporal horns.

March 2004 – So many people have e-mailed me on saying that my husband MRI results are just like there love ones.

July 2004 – Steve is so tired during the day. He has to take a daily nap. He cannot multitask as before. It is hard for him to focus.

Nov 3rd, 2004 – Steve he felt like things were moving in front of him had a pain scale of 5
He felt intoxicated and had slurred speech – took aspirin, had pain above his left eye – chalk white – did not look good Had a TIA after the TIA he had a four hour nap. They say that when you sleep the brain tries to heal.

Steve refused to go to the Emergency Room but I insisted. Within an hour at the E.R. they sent Steve home, as there was nothing they could do.

November 2004 – Steve's memory is going. He would say, did you feed the _____ you know what? Then point to the object. Saw the Directors of stroke and suggested maybe putting Steve on "Aricept" to see if this helps. A 48 year old man with Alzheimer's disease can you imagine that. Within two weeks, the Aricept seems to be working his memory has improved.

January, 2005: Dr. Pastores of New York University School of Medicine called me to see if anyone I would be interested in getting patients to participate. The study would be for short term memory loss and that it was for a medicine for Alzheimer's disease. I asked him if it was Aricept, and he asked how I knew. I explained that Steve was put on the medicine last month due to his memory loss. I said I would be happy to assist with getting participants. This was the first ever study for CADASIL Patients. The purpose of this study is to determine the safety, tolerability, effectiveness, and side effects of an investigational drug compared to that of

placebo in the treatment of subjects with mental decline associated with CADASIL. The diagnosis of CADASIL is established upon brain MRI findings consistent with CADASIL, and: 1) Identification of a NOTCH 3 disease mutation, or 2) the presence of deposits (on electron microscopy of a skin biopsy). Patients with CADASIL, who are between the ages of 25 and 70 years, and have cognitive impairment, are eligible to participate. The study will take place over 18 weeks.

January 2005: I was invited to attend a CADASIL conference on January 29, 2005 at Newcastle-upon Tyne, in England. I had the opportunity to attend the CADASIL Awareness Conference/Symposium (First of its kind in the World). This is more than I could wish for. I feel like I am in a movie and waiting for someone to tell me "cut, it's over". I talked with Doctors, Professors, Scientists, CADASIL patients, families of patients, etc. (little old me from Texas). The conference/symposium was wonderful. Everyone knew of, or had heard of my name, Billie, through my website. I was amazed, how one person could be known so much by so many.

I met at least 3 families that I had been e-mailing about CADASIL and had assisted them from the beginning. One patient: whom I had emailed personally the week before the conference/symposium without knowing I was going. The gentleman came with his sister and could not believe I was there. I met two daughters, their mother a CADASIL sufferer whose father was an advocate for his wife. He had contacted me about seven years ago to find out what to do. It is a small world after all. During the evening meal I sat with the above families at the same table and a world expert, Dr. Martin Dichgans from Germany. Dr. Martin Dichgans became interested with CADASIL right after Medical School and has not stopped since. I couldn't eat as I was so excited to meet everyone and felt much honored to be there. Jack Shields, who was the one who invited me to come, has CADASIL, and he was treating himself with a supplement called L-Arginine. Jack Shields was the gentlemen who was in charge of the conference and had established the CADASIL Trust in England. He gave L-Arginine to me to take back home for Steve to try. I was sure it would not work as I have heard of all the treatments that others have tried. Steve started to take the L-Arginine supplements the night I arrived home. Within 1 month, and now over 6 months later his quality life has improved tremendously.

February 2005: I finally completed my Associates Degree in Computer Technology Management, which I worked on for 2
years.

I received an autopsy report from a wife whose husband had CADASIL. She has 3 other children, two of which have CADASIL. I scanned the report and placed it on the website under Professional Health Pages, so hopefully this will help doctors understand CADASIL.

March 2005 – Steve has been taking L-Arginine for 2 months now. He does not walk with a cane anymore and feels like he has more energy and no headaches at all.

March 2005: Over the spring break holiday, my sister and husband encouraged me to start a foundation here in the USA. I bought the book, "Nonprofit Organization for Dummies", and have not stopped since. The Director of Stroke who Steve sees wrote- "Keep up the wonderful work you are doing. It is a real help to other families with CADASIL".

May 4, 2005 – I had e-mail from a lady who father has CADASIL who works for the BBC in Ireland. I introduced her to Jack Shields and he asked her to be on the trust in England. She is trying to get the story out about CADASIL. She had asked me to complete the questions, which are below. BBC asked: I will like to state those CADASIL sufferers and their families received very little information and help from members of the medical profession. This certainly is the case concerning my father and many others I have met through the support yahoo group in the USA. I understand CADASIL is a new illness but I honestly feel more can be done on their part. Do you agree?

Billie and Steve say: We agree more should be done.
Steve states: I did not go on the USA yahoo groups as it gets very frustrating when people are feeling sorry for themselves, etc. My wife Billie started the journey for me 8 years ago when I was first diagnosed with white matter disease. She initially contacted anyone who would listen, scientist and doctors who were or had some knowledge. She has through her website connected to hundreds of patients and families and doctors in most parts of the civilized world. The doctors are working very hard towards a treatment and hopefully a cure but still little is known about CADASIL.
Billie states: I started the website and Doreen spun the USA yahoo groups of the website a couple of years later. I used to get upset with the yahoo groups especially when they scare others about CADASIL. It has good points and bad points to it. I use to cry with the answers of the yahoo groups,

as I was scared this is how Steve will be like. (Not true) There are no good answers yet to CADASIL and people try there best and yet it gets very upsetting reading info that is not correct.

I have helped at so many people get diagnosed with CADASIL, The medical profession who know about CADASIL tell patients or hand out my literature or tell others to get on the Internet and go to my site as the information is there.

BBC ASKED: CADASIL is hugely underestimated...There has been cases misdiagnosed as MS. I know of a few cases here in the UK. Is it the same in the US?

Steve state- Yes, most definitely most neurologist has never heard of this disease so they look at the first most one that has a few similar symptoms.

Billie states: Yes, it is underestimated all around the world. Since I have the guest book on the website I have over 276 confirmed cases of CADASIL all around the world and having different people sign in at least 5 times a week with new patients or people or doctors who want to know about CADASIL. At present these are the figures : 200,530 hits last year alone 2004

my question is why so many hits? Are people looking for cures or even answers to CADASIL? And together there is hope.

BBC ASK: Being ones own cases manager' and taking the initiative to do your own research has proven to be very successful. How else would people know that 'they are not alone' and 'together we have hope'? The Internet is an absolute godsend as it is the medium you can gain a better insight into the illness. Many people (not just CADASIL sufferers) take what their doctor / neurologists say as 'gospel', I firmly believe that if my family simply listened to the doctor's advice my father would be dead today

Steve states: Doctors because of their training take what they see and read in medical journal as the gospel. They only rely on evidence and proven facts finding the right doctor is very key one who is willing to allow take safe risk and try alternate methods. Doctors in the UK are high and mighty in the USA they are more realistic and listen to the patient most the time.

Billie states: from the very first I have suggested that you have to be your own case manager or otherwise it would not help you love one. I do not know if I would have Steve now if only through my insistence on take control of his care and informing doctors. I was brought up to believe doctors are the answer to our prayers. It was hard for me to understand they do not know everything. I have encouraged patients and families to inform and educate their own doctors and don't give up on them. If the doctor does not want to be educated locate another doctor who will. My own expense I mail out brochures, medical information for patients to give to their doctors to educate them. I even mail out the information directly to doctors and some doctors even has called me very interested in CADASIL and thanking me personally for taking the time to inform them. I have doctor e-mailing me on my efforts and what a wonderful resource the website has been for them. Patients has told me the doctors has told them to go to the Internet and look up my website. I get e-mails asking for help!

BBC: Finally I really believe that Steve's progress has been amazing. I would be really grateful if you could do one more thing for me?!...Will you give me a brief outline of how Steve was on a daily basis when he was at his worst with CADASIL? My father's condition has also certainly improved and he has only been taking L-Arginine for 3 weeks!!

Steve states: Prior to L-Arginine, I had trouble with motor functions (walking getting out of chairs) Blinding headaches vision disturbances, flashing lights, etc. single tasked (one thing at a time) Confusion with thinking skills, poor sleeping
could not swallow well, slurred speech when tired, pains in arms, trouble with trying to explain simple things, poor working memory TIA's/strokes, tingling in fingers/toes/faces drooling, high pulse rate, All of

my above mentioned symptoms has subsided greatly or vanished since taking L-Arginine in January 2005. I take 1 12 gram per day (6 grams in morn and 6 grams in evening with water and blended very well.

Billie's states: Prior to L-Arginine, Steve was not functionally very well and now he has fixed the kitchen sink. He still needs to take his daily naps but otherwise he seems more alert. I really thought that the L-Arginine would not work but I ate my words as with Steve's other medicines they all help out. When I am on the computer and hit the send button on e-mail that one day there will be a cure or a treatment from this nightmare. When I started to research CADASIL, how I wished something would happen to make others be aware of CADASIL, which included a treatment or cure. I have been an advocate all the way. Telling everyone to be there own case managers and suggesting to print out the information from this website and inform your doctors or who would listen. I receive e-mails from doctors, patients, family members, etc. The worldwide support of the website has been unbelievable and no words could express the appreciation to everyone. Each month e-mails were increasing and phone call increased. And on May 10th, 2005 CADASIL Together We Have Hope became a nonprofit organization (www.CADASILfoundation.org

May 10, 2005: I completed the Articles of Incorporation for CADASIL Together We Have Hope Nonprofit Organization, and filed them with the State of Texas. When I started to research CADASIL, how I wished something would happen to make others be aware of this disease, which included a treatment or cure. I have been an advocate all the way, telling everyone to be their own case managers. I suggested they print out the information from this website and inform their doctors or whoever would listen to them. I receive e-mails from doctors, patients, family members, etc. The worldwide support of the website has been unbelievable, and no words could express my appreciation to everyone. Each month, phone calls and e-mails were increasing. On May 10th, 2005, CADASIL Together We Have Hope became a nonprofit organization (www.CADASILfoundation.org)

May 6, 2005: A research Doctor from Germany e-mailed me- "Billie thanks for your e-mail. Of course I remember you very well, and I was happy to meet you in England in January. Thank you also for your kind support. You are doing a great job. I can see that you are extremely busy, and I told you that I think it is very important for you to have some time for yourself outside the CADASIL topic. Let us hope that we can make some progress in the next few years. Kind regards".

May 2005: Dear Billie- You have been an inspiration to us all! Dr. Gregory Pastores, Dr. Charles Zaroff, and I at the NYU Neurogenetics Clinic has been seeing a number of CADASIL patients referred to us by you and the ULF. Several are now in our clinical trial. I look forward to your participation in the July ULF annual meeting.

May 10, 2005: Received the Certificate from the State of Texas for Incorporation of the CADASIL Together We Have Hope Nonprofit Organization.

May 16, 2005: Received the first donation to CADASIL Together We Have Hope, opened account. I contacted testing sites for CADASIL all around the world (part of our mission) and asked if I could let other know about how to get tested. I had wonderful response.

May 25, 2005: To support the CADASIL patients and their families who visit your website and refer to it, we will grant a reduction of 10% on our price for CADASIL testing. One could encourage these persons to donate this amount to your organization. It is my strong opinion that such patient support groups help the affected and their families by supplying information on the latest scientific developments concerning diagnosis and treatment. What is probably the most
Important, support groups like yours provide a platform for exchanging every day problems and solutions in dealing with this disease, which strikes like lightning and changes the life of the affected family completely. It also will give the patients the feeling that they are not alone with this rare disease. This gives strength and hope! A well informed patient group can also improve the awareness of medical professionals. Because diseases such as CADASIL are so rare, they often are missing in the education plans of medical students. This leads to doctors that have never heard of some diseases and patients seeking advice going through an odyssey to get their symptoms interpreted right. Thank you for putting our address on your website (from Director Gene Analysis Service GmbH Grainauer Str. 12, 10777 Berlin, Germany).

May 26, 2005: Human Genetics contacted me and said congratulations on your spectacular web site for CADASIL, and to let everyone know at Boston University School of Medicine has made history in achieving the

first ever prenatal diagnosis of CADASIL. The scientific paper reporting this achievement has been submitted for publication. Call 671:638:7083 for more information.

June 2005: Newsletters were mailed out to over 300 addresses worldwide announcing the organization, the Study, and to let others know about L-Arginine.

June 2005: Completed the IRS Paperwork to file the 501 c 3 forms.

June 20, 2005: I was notified that an article from the Charlotte Observer was posted on "Family Tree Loses Genetic Defect". See the website- http://home.earthlink.net/~CADASIL/News%20articles.htm

June 20, 2005: Contacted the writer of the article and she responded: "You are welcome to post my article on your web site if you just give credit to the Charlotte Observer and me. At this time, I probably won't be doing another article on the condition, but I'm glad I was able to bring some awareness to it through the story about Toni and David Brinson. Thanks for your email. 'm glad to hear from you, and thank you for taking time to write". Karen.

June 21, 2005: Contacted Dr. Mark Hughes, director of Genesis Genetics Institute in Detroit who was mentioned in the article and this is what he said: "Thanks for your note. Indeed, CADASIL is a dreadful disease. We are happy to be helping couples remove it from their family tree forever, and do so before they ever begin a pregnancy. One thing too.... the cost for IVF varies GREATLY across the nation. So, putting a cost of $25,000 is valid for some centers on the east coast and in LA, but it costs significantly less ($12,000) in much of Middle America. The actual Preimplantation Genetic Testing is done for less than cost at $2,500. Most of the actual costs come from a gift foundation. Keep up the great work! Families need organizations and information websites like this! Congratulations. Best regards."

July 2005: Attended the United Leukodystrophy Foundation conference the week of July 20 to July 24. It was a wonderful experience. I have been enrolled with the ULF for over seven years now. There were wonderful researchers, neurologists, and scientists, which were present at the conference. Some knew about CADASIL, some did not. I distributed the "Together We Have Hope" cards to everyone. These professionals were so supportive of what "CADASIL Together We Have Hope" (Foundation) is doing. I even had an Associate Professor of Neurology and Pediatrics, Neurogenetics Laboratory, Department of Neurology volunteer to be on the scientific advisory board for the foundation. We already have been working together since January. This professor is establishing the Scientific Advisory Board and we have approached by a number of U.S and International Investigators who may want to be a part of this wonderful effort. I met a couple whose daughter has MLD and had started a foundation. It was explained that one of the goals of the organization was to have a domain site. They volunteered to set up www.cadsailfoundation.org.

It is very important to know the mutation of your gene that is affected with CADASIL. This will help generations in the future to be tested for CADASIL, as only a DNA blood test is needed. The doctors will only look for the defected mutation. There are 50 mutations in CADASIL, which is high. This is the reason why it is so hard to find a treatment for CADASIL. The mutation can be inherited from one generation to the next. I talked with Genetics' about my family.

I finally got to personally thank Dr. Hugo Moser and his wife, the doctor who had taken my husbands MRI and his blood to assist in diagnosing Steve. I met Raphael Schiffman (research/doctor) who had called me in 1996 to tell me that he thought my husband might have CADASIL. We had e-mailed back and forth, and I finally got to meet him to personally thank him for his help. I did ask him what he was working on now, and he said a rare disease called Fabry's. I could not believe this as a friend of my husband was diagnosed only 2 weeks before the conference with this. He gave me his business card to give to them.

I was eager to meet, a person from Nevada who has CADASIL, as I had assisted her about two years ago. During the conference, a gentleman from Ohio had called my house and wanted to talk with me about CADASIL. My husband had explained that I was at the conference in Illinois. He said he was leaving tonight to go to that conference, since he was registered. You could say we had our first unofficial meeting in the USA for CADASIL with the people from Ohio, Nevada, a lady who just was tested (her brother had CADASIL), and myself. We gathered around a table and talked for over 90 minutes. We talked about everything including the "CADASIL Together We Have Hope Organization".

During the presentation on CADASIL, the presenting professor mentioned my name several times saying that "Billie" has assisted with CADASIL or "Billie" can answer that question. My 17 year old daughter wants to be tested for CADASIL. I am so scared to know. My 21 year old does not want to know. The conclusion for our family is to find out what mutation my husband has, since there was not a blood test available at the time of

diagnosis. When we know the mutation, we will locate a genetic counselor that knows about CADASIL. It will be my children's' choice on whether to get tested now or later.

CADASIL is passed from parent to child through a mutation in a gene. Each offspring of a CADASIL parent has a 50% chance of inheriting the disease. If a person does not inherit the CADASIL gene, he or she will not develop the disease and cannot pass it to subsequent generations. A person who inherits the CADASIL gene will sooner or later develop the disease. We have the dilemma of whether our CADASIL offspring should or should not be tested for CADASIL. There is a good article at http://www.hdfoundation.org/testread/russroul.html, Genetic "Russian Roulette" The Experience of Being "At Risk" for Huntington's disease. Huntington's Disease has a 50% chance of inheriting the disease like CADASIL.

When returning from the conference, I e-mailed everyone I had met that "it was a pleasure to meet you at the ULF conference last weekend. I was so happy to talk to you concerning my husband." Steve was diagnosed with CADASIL in 1997. I have not stopped networking since 1996. I have two daughters who have a 50 percent chance of having CADASIL.

On the foundation database, there are over 264 confirmed cases that were diagnosed through skin biopsy or blood tests. If you go to this link, I have a map of all confirmed cases all over the world marked with pink squares. As of July 2005 http://home.earthlink.net/~CADASIL101/tracking/

There are at least 60 doctors who have seen at least one patient with CADASIL, so if a person calls requesting a doctor's name, we can refer him or her to a doctor. When attending the ULF conference, I did notice I was already following the guidelines for HIPPA. The response from everyone was so supportive.

July 31, 2005: A doctor named David whom I met at the conference wrote, "You and the CADASIL Foundation are firmly ensconced in my Palm Pilot, and I will certainly pass the name of your foundation on when the opportunity arises. Please let me know if I can be of any help."

I posted the PowerPoint presentation from the conference I attended on the website under Health Care Professionals. The doctor had forward me the presentation. I posted MRI and skin biopsy reports also. The registrar with Genetic Alliance as CADASIL was not recognized with the organization.

Aug 3, 2005: We need to have the American Stroke Association recognize CADASIL. I sent the American Stroke Association an information package, and they responded with this e-mail: "Thank you for writing the American Stroke Association and sending information on CADASIL along with your family's history. I apologize for the delay in responding to you. I have forwarded your information to our Stroke Connection Magazine staff for consideration for possible inclusion in the magazine. The Stroke Connection Magazine staff reviews new material for the magazine every two months.

August 4, 2005: A doctor had e-mailed: "It was wonderful to meet you in person, and your drive and energy is inspiring. I hope the CADASIL families appreciate what you have done and are doing for the group."

August 5, 2005: "Thanks for all your energy and hard work. It is appreciated all over the world!" Regards Sonja (South Africa)

August 6, 2005: A gentleman called me to let me know about an article in the Men's Health Magazine about CADASIL, "Hunting My Father's Killer". It was a detailed article about CADASIL. Sunday, I e-mailed the editor to pass on the e-mail to the writer. On the same day the writer of the article e-mailed: "Thanks for your kind letter. My heart goes out to you and your family. I'd come across your web site while I was researching my story, and found it very informative. It's amazing to me that this disease has gotten so little media attention: Men's Health is probably the first major magazine to address it. I may have the opportunity to do a short follow-up in an upcoming issue. If it happens, I'll definitely mention your web site. Also, I believe we are putting the article on our web site, http://www.menshealth.com we can link to your site from there as well. If you're interested, I could provide a .pdf of the story for you to post on your site. I too want to raise awareness of this disease, and I'd like to share my story with the CADASIL community. Let me know if you're interested. Let's stay in touch.
Thursday, August 12, 2005: I posted on the website the story at, I e-mailed this out to all the people on the database, and by Sunday, August 13, there have been 284 people who reviewed this article.

August 13, 2005: I received this e-mail: "Wow ::: very interesting article. Thank you for keeping me updated ::: I truly appreciate it".

August 14, 2005: Search on database for CADASIL CHID is a bibliographic database produced by health: related agencies of the Federal Government. This database provides titles, abstracts, and availability information for health information and health education resources. They had CADASIL : Together We Have Hope on their databases. At the end of September, I will be representing CADASIL at a National Organization of Rare Diseases meeting in Rockville, Maryland. This conference is a collaborative effort of the National Institutes of Health, the Centers for Disease Control and Prevention, the Health Resources and Services Administration, the American Society for Human Genetics, the American College of Medical Genetics, the Genetic Alliance, the Society for Inherited Metabolic Disorders, the National Organization for Rare Disorders, and Emory University Department of Human Genetics to address the growing public need for improvement in availability, accessibility, and quality of genetic and other diagnostic laboratory testing for rare diseases and conditions. I Received an invitation from N.O.R.D to attend this conference. I received a phone call from CDR, US Public Health Service Director, Extramural Research Program, Office of Rare Diseases, OD, NIH, and 6100 Executive Blvd. Rm. 3B01:MSC 7518, Bethesda, MD 20892:7518 asking my husband if he would consider being a patient advocate at the meeting. My husband mentioned that I was going, and she said she had already heard of my name, "Billie". I Created a chocolate bar wrapper on the computer to raise awareness all over the world.

August 21, 2005: I received this e-mail: "Wow, this thing with the small candy bars is going crazy here in Oak Harbor, Washington. I have friends that have already taken them off my hands, so I was wondering if you have a program that you could attach to an e-mail so I can print them at my house.

Steve is functionally better than he was in previous years due to all the 13 daily prevention medicines he is on. He does not live in pain. He still gets very tired, has short-term memory loss, sometimes has a short temper, and is easily fatigued. There is no way I can end this story, I am here for my family and everyone who has this disease. You see, when my husband dies, this disease does not die with him. I love my family! Daily, we receive e-mails or phone calls for assistance or just to talk. These are our goals and what our foundation will be working towards. We are in the first stages of considering writing a grant.

Steve is functionally better than he was in previous years due to the 13 daily preventative medicines he is on. He does not live in pain; however, he still gets very tired, has short-term memory loss, and sometimes has a short temper sometimes). There is no way I can end this story, I am here for my family and everyone who has this disease. You see, when my husband dies, this disease does not die with him. We receive daily e-mails or phone calls for assistance
Or just to talk.

CADASIL TOGETHER WE HAVE HOPE: Planned Activities

- To promote awareness.
- Understanding of the condition, its symptoms and possible sources of medical assistance including knowledgeable physicians, qualified labs and updated research information. Physicians and the most of the medical communities are not aware of this rare disease. Patients are frequently diagnosed with Multiple Scoliosis or an undiagnosed Leukodystrophy (white matter disease.
- CADASIL is rare, genetic disease; usually affected families remain unaware of the presence of the gene and may suffer the loss of one or more family members. We intend to undertake a variety of public awareness efforts through general and mass media and through publications targeted at the medical community.
- Fundraising events will play a part in our effort to increase general awareness and understanding of CADASIL.
- To educate and support doctors, patients and the public.
- Educate doctors, radiologists, neurologists, and directors of strokes, and others who may well be presented with a person with symptoms of CADASIL, but fail to recognize them as such.
- By updating the internet website with CADASIL information, CADASIL studies, testing sites, news articles, newsletters, research, doctor pages and confirmed cases.
- Reporting the outcome of the attended conferences.
- Mailing out information packets to doctors, sufferers, etc.

Other activities:

• Work with National Organization of Rare Disease (NORD) in developing clearinghouse for low cost genetic testing
And diagnosis.
• Secure physicians and neurologist to be patrons of the organization.
• Secure additional qualified board members.
• Develop and hold symposiums on CADASIL in the U.S.A.
• Develop goals for the organization and review each year to assure proper focus of the group.

Samples of CADASIL Related E-Mails From Around the World – Unedited

1998: A 39:year:old father has CADASIL. For years, the doctors thought it was MS. He had a major stroke three years ago, which made them look further into his complications. He was not able to eat, talk, swallow, walk, or hardly ever communicate. Two of his three brothers have also been diagnosed with CADASIL. They suspect that CADASIL is what his father died from. He also has a niece with the disease. He has two young children who could have CADASIL.

1998: A man who is 50 years old has been diagnosed with CADASIL. He has numbness in his feet. He was a police officer for more than 10 years. He lives in the United Kingdom.

1998: A wife called to say her 52 year old husband had been diagnosed with CADASIL (Sept 97). The doctors thought it was MS at first. He has fatigue, short term memory loss, lack of concentration, and has gait changes. He is on disability, as he cannot work. His father died at 64 years old of stroke, with lost of memory around 58 years old. His grandfather died of a stroke at 63 years old, and his great-grandmother died of a stroke at 54 years old. He had a sister who also died at 26 years of age.

1999: Hope things are ok with Steve. B has not been doing well at all. He cannot walk unaided at all now and cannot even do the easiest of tasks by himself. We have been working hard at trying to get some help for him during the day while I am at work. I have exhausted all county help, and now I am trying to fight the insurance company to provide some home health aids. It's not bad enough you have to deal with these physical and mental problems, but to have to deal with all this other stuff just adds to the stress. Did you see the Christopher Reeves documentary on Nightline last week? It really spoke too many of the problems we are experiencing even though he has a different problem. A lot of what you go through is the same. I am going to try to sit down today and start telling my husband's story. I have been keeping notes, but they are on this paper and that paper. If and when I get this done (in my spare time: yea right) I will mail you a copy and if you want to put it in a newsletter you can. Just wanted to touch base. Even though we don't communicate much, it's nice to know there is someone out there who understands what you are going through. Really do hope things are ok with you.

1999: Recently a Registrar, at the hospital my mother goes to, thanked my father for updating him about CADASIL and
News about the research now being done in the UK! My father has sent details of this site to the consultant who is in charge of research in NEWCASTLE (he knew of Billie and Steve's site).

1999: I have just discovered your web site, using my son's computer. My wife, aged 62, and two daughters, aged 37 and 34, have all been diagnosed with CADASIL. They diagnosed my wife and eldest daughter after having MRI scans, and my youngest daughter after an MRI and a skin biopsy. She insisted on these since she has two young children, and is obviously concerned that they may pass it onto them as it is an Autosomnal dominant condition. You are probably aware that there is little, if any, research being done in the UK. I have made contact with a Doctor in Munich who now has DNA samples from all family members and is checking it to try to find the mutated gene on chromosome 19, which they think to be responsible for the disease. I have written to 20 health authorities in the UK, and of replies I have received, the total of recorded cases was only 12. I am anxious to find out how many cases have been confirmed in the UK, but getting information is very difficult due to patient privacy laws here. You are also probably aware that the disease is largely under: diagnosed, and there are many thousands of people who have CADASIL, but because GP's are not aware of CADASIL, they are not referring patients to neurologists for tests. Do you have any suggestions how to raise public awareness, as this is the only course that will lead to more research and hopefully treatment and cure? I have been doing as much research as I can. I use companies such as MEDLINE and using Radiophone in questions where I have managed to get CADASIL discussed on UK national radio for the first time (FEB. 1998). It's nice to see someone else out their being positive and trying to raise the profile of this distressing disease.

2000: Male 38 years old. Could you please send the CADASIL newsletter? I have suffered a stroke on the right side. After numerous tests, it was discovered that I have CADASIL. I want to know all the info that is out there and will help in any way possible. I have children aged 5 and 8 and would like to be around awhile. Thanks

2000: Person 41 years old. Last year I was diagnosed with CADASIL. T has been a long journey for me, and now I am having so many problems getting my medication for this. The doctor says I have so many lesions to the left side of my brain, and there is so little information about this disease. I was hoping you could show me some new information.

2000: First let me thank you for establishing your web site: searching for Information about CADASIL has been difficult. I know two years ago there was very little, if anything, about this disease on the Internet. My father's family has been severely affected by the disease. In fact 4 out of 8 of his siblings have been diagnosed with it

(thus far: three remain to be tested). Although my father shows no signs of the illness at age 45, my sister and I know he is not in the clear yet. Please add my name and email address to your contact list. I am a young university student studying Neurology/Biological Psychology. My interest in CADASIL is both professional and personal. Thank you. Let us hope one day soon there will be a cure.

2000: From France- I'm writing from Paris France, where I live. I'm not sure of my English medical vocabulary, but I will try to tell you my story. My father has CADASIL and has been diagnosed in 1995 after a cerebral infract which lead him to a hemiparesy. He has not recovered yet from all. He is having also a small depression and some mood problem. He is followed by the team of Pr. Bousser and Dr Chabriat at the Lariboisière Hospital in Paris. I have had a genetic test to know about my case, and I'm waiting for the results next February. I've had no symptoms since now. I'm also working on a French site for CADASIL with an association of patients of Pr. Bousser: ACF. First we'll do a French version and an English version is planned too. I've not read all the archive of Onelist, but I will be pleased to give you all the details I know. Together we have hope.

2001: My daddy is 59 years old. He has been diagnosed with CADASIL. We have struggled for answers since 1991 when he became depressed, incoherent. His physician diagnosed depression and prescribed Prozac, which he took briefly and then "returned to normal". We did not have any problems from 1991 until 1997. He was well except for "ministrokes" and "white matter" diagnosed all in 1997. Since that time, there have been so many behaviors, symptoms and changes that did not add up. Our neurologist told us to live with it...there was nothing we could do. We initiated a trip to
Research Hospital in Gainesville, FL and found out 10/4/00 that Daddy has CADASIL. I am 28 and my sister is 31. There are three grandchildren. My sister and I have not told my parents that this horrible disease is genetic, and we probably never will. I was told to give him 2000 units of Vitamin E daily and 100 mg of Zoloft. He has problems swallowing, speaking and shuffles when he walks. He has never had migraines or headaches. He is not in any pain at present. We have very little family medical history. We have traced the disease to my Daddy's mother whose father was placed in the
State Hospital (early 1900's). Daddy's mother was in a car wreck at age 56 and stayed in the hospital for @ 4 months. She was a vibrant, active and very outgoing lady from the stories I have heard. Especially so when she was hospitalized for those 4 months. However, the story goes that she was told by the hospital that she was not going to get to go home because her leg had not healed. According to the history, she simply died shortly thereafter. No symptoms, no mini-strokes, and no personality changes at age 56. Could this mean that if you are healthy you may not develop the disease? Also, does the disease weaken with every generation? Sorry to ramble and bombard you with so many questions. It sounds as if my daddy has it pretty good as he has no pain at all like your husband Steve. Thank you for this web site and your dedication. Please send information if you have any.
2001: Hello, I found your web site just now and decided to contact you for??????? My lovely 41: year: old daughter was given this diagnosis about 2 hours ago and we are in a state of shock. The docs have been toying with MS but one doc held out and said no. So this is where we are.

2001: I want to thank you for the tremendous research you undertook to compile the questions and answers on CADASIL. I just returned from seeing my neurologist to get the results of an MRI of my head and an MR angiogram. He informed me that the results suggest that I have CADASIL. Both of my parents died of strokes and my brother has had a series of strokes, the first of which was 10 years ago when he was 45; all of which makes him think that CADASIL could explain what recent tests have shown. I have been having headaches for the past 4:5 months, which prompted a CT scan in July 2000. The CT scan report noted, "There is a subtle area of hypo density seen within the subcortical white matter of the right anterior frontal lobe". In 1995, I started having two disturbing problems: a) memory problems and difficulty in concentration; b) I had developed a tremor which prevented me from writing or holding things without trembling (the tremor is still with me, and is often worse; fortunately, I am able to type, and the tremor does not affect my typing). To explore these problems, the neurologist at that time ordered an MRI angiogram, which has served as a baseline to compare the MRI and MR angiogram that was carried out in October 2000. In the earlier test, the report noted signs of atrophy in the white matter. The latest report shows a significant deterioration and damage to the white matter, and indications of lack of blood circulation in the area. I have no history or diabetes, high blood pressure or high cholesterol, and I do not smoke. However, my doctor says that the extent of the damage is similar to someone with years of diabetes, high blood pressure, high cholesterol, and a heavy smoker. Therefore, considering my family history and the results of the latest MRI compared with the earlier one, he has made a preliminary diagnosis of CADASIL. He has referred me to a stroke specialist to run a series of tests to try to confirm this. I see him in a month. I have a few questions and I wonder if you could share your thoughts. I appreciate that you are not a medical practitioner, so I am not asking you for medical advice, only your experience based on someone who has been so intimately involved with the disease.

2001: Dear CADASIL newsgroup's members and readers. I am a French person who feels very much involved in the CADASIL disease's cause. This illness was discovered in my family. Many relatives took part in the

medical research, which was launched in France more than 15 years ago and led to the identification of the genetic disorder at the origin of it, in 1993. My grandmother died of it, as several of her brothers and sisters, and the genetic disease has unfortunately spread among the following generations. In France, about 100 families have been identified as having cases of CADASIL in their genealogical tree. This means that many people suffer from it, or are afraid of seeing their health situation worsen, or are afraid of transmitting it to their children, or are afraid of having got the genetic abnormality from their parents, or live with persons suffering from it and strive to give them medical and moral support, and so on. You live in different countries, but the same feelings arise in the messages you exchange through the newsgroup. There are no boundaries in our concerns with CADASIL. It is quite necessary to find understanding and solidarity when such a rare illness invades your life, but, with an exterior view, I find very few hopeful or scientific proven messages in your newsgroup. Do you have some associations for sharing your feelings, exchanging advice for caring of ill people and getting medical information? In France, some scientists work on this illness, but each of us or our doctors cannot individually contact them to get information. An association can be very helpful for these researchers to have a way of conveying information and for us, as a group, to get real and positive reasons for knowledge and hope. Here, in France, we founded an association, "ACF France- Arteriopathy Cérébrales Familiales France", which means familial cerebral arteriopathies, France. One of our main objectives is exchanging information with the French research team and communicating it to our members, and also to medical media, neurologists, hospital teams, etc. In that purpose, we are preparing a Website. We will give you its address, but it will first be in French, but we plan to translate it in English later. We will prepare a newsletter, get some contacts with media, and give support to families. Our association has already 90 members and has also subscribed to another French association dealing with rare genetic diseases. Our first annual meeting will be held in the coming weeks, and it will be followed by a medical conference. Together, we will be stronger! There's a French Website where you can find some extracts of medical articles on CADASIL, in English. Its address is http://www.infobiogen.fr/services/orphanet Choose "search the orphaned database", then "disease", enter CADASIL and select [search], click then on the CADASIL name and a list of more than one hundred articles is displayed. You can click on their names for getting options like displaying abstracts. I hope we can help you in knowing better the CADASIL illness and having information on the prospects for medical research. Yours sincerely,

2002: I've been in denial over the fact I have CADASIL. The depressions are the worst part of it and the fact that I forget a lot of things. Believe me, I write a lot of notes for myself and sometimes forget where I put them. The only thing the doctor has me on is Coumadin 9mg and Tylenol with codeine for pain. I'm looking for help in the memory department. I have found diet and exercise helps me. I know it sounds dumb, but it works. Any help is welcome. Dave

2002: My doctor is out of Cape Girardeau. I also take Depakote for my migraines. I take Pail for depression and anxiety. My father, sister and youngest brother went to Mayo Clinic in Rochester Minnesota for testing. My other brother and I went to Columbia, the University. My doctor up there is Dr. H. My brother is in St. Louis and was in the Barnes Jewish Hospital for two weeks for three strokes. They have moved him to their rehab facility. It looks like he might be there another three weeks. There is a doctor there that knows about CADASIL, and he sees two other families. My brother is unable to walk, dial a phone or write. He is like a child, and he is only 34 yrs old. I hope this helps.

2002: My husband has had strokes since age 35. He is now 49. Eight years ago this month, he had the BIG ONE. Like you, I mourned the man I married. I lost my husband, my lover, and my best friend. And yet he lives. He is totally paralyzed on the right side now, and his personality is completely different. He is very child like. But just when I don't think I can stand any more, I see something in his eyes that tells me he is in there...somewhere. I have him at home with me. He is too young to be in a nursing home. I will not allow that to happen to him too. I struggle every day to make ends meet and I know it is worth it. You see, like you, I love my husband. I loved him before he had symptoms, I loved him when he was grouchy, when he had trouble seeing, when he would stand up and then just fall, when he slept for 16 hours or more straight, when he couldn't sleep for days, when he cried and couldn't stop, or laughed and couldn't stop. I loved him when his memory got so bad that he didn't know who I was or where he was. I loved him when he couldn't talk and had to be fed through a PEG tube in his stomach. I love him still. He is still my life. Most of my friends don't understand how I can stand to take care of him always. Some have told me to put him in the nursing home. They do not understand that his place is with me. That is my choice, my decision. There may come a day when it will be necessary, but not today. Today he is still mine. Maybe that sounds sort of sick, but I don't mean it that way. I have mourned my husband, and yet he lives! This is a condition that most people don't have to deal with until they are old or they need to care for older parents. But CADASIL has given many of us this reality. Remember the spouses and family members, too. My husband doesn't realize what he has lost any more, but I do. So, love them while we can. Live for this moment. Enjoy holding hands, simple conversations, and shared moments of simple silence. They are what keep me going, now that I no longer have them. I am 44 years old and my husband is still with me. That much I am thankful for. Kay

2003: I'm a 54: year old male who has just recently been diagnosed as having CADASIL. My doctors found a little white cloud around my brain on an MRI. I would like to know how fast do the symptoms progress? I already have trouble walking in tandem (heal to toe), can't ride my bike, and unable to ski. These are things I could do just fine a year ago. What can I expect next? Thanks.

2003: Hi, my name is Chris. Thank you for putting up the Egroups (now called Yahoo Groups) site. Thank you Billie for your site and for answering my first e-mail. I will try to keep this short. My wife is 40. My wife spent this past Wednesday in the ER. Her symptoms were merely tingling in the arm, but her family had a history of strokes so we went in. Of course, after several hours they reassured us everything was ok. The next day she went to her MD because everything was the same. On Friday, she had the tingling and a throbbing in her neck. Although our MD reassured us that it wasn't a stroke, she insisted she have a cat scan. The CAT scan showed something, and she was immediately taken for a MRI. I wasn't even there for the first test because the doc said everything was ok. I showed up as the MRI was performed. At some point, I'm sure I will have to deal with that decision, and my wife knows that I will be at EVERY doctor's appointment from now on. The person who read the MRI said CADASIL. My doctor and the neurologist were clueless and had to look it up. I had to call my father-in-law to get any information on his wife. They had had the genetic testing. She had it, but we did not know. Her grandmother had strokes and her great grandmother had had strokes, but nobody knew why back then. Thank you again for this site. I consider myself fortunate at this point because this is my wife's only problem at this point. The MRI shows that she has had several TIAs beginning in her 30s. At this point in time, we know there will be more. Billie, I have already learned why you told me to be my wife's case manager as I have already found connections that my wife's doctor probably would never have found. To one and all, you MUST be their advocate as well. It was my wife's insistence that led to her first test.

2003: CADASIL is wiping my family out! Almost everyone on my mother's side is lucky if they make it into their late fifties. Many died in their forties. I am forty five and am not doing too well. I am a bit scared because my daughter started the CADASIL symptoms at the age of sixteen. She also has the other illnesses that I have, which are fibromyalgia and chronic fatigue and immune system disorder.

2004: My husband is tired all the time. He sleeps an average of 15 hours per day. If he doesn't get his sleep, he doesn't seem to be able to cope as well, and his thinking is not as clear, so I don't complain and just let him sleep. I am alone a lot.

2004: My husband does have a minimum of 2:3 strokes each week. Some last a few minutes and leave him drained so that he must sleep for a few hours. Others may last throughout the day and possibly the next with lots of sleep needed. Thank heaven he is motivated to do lots of home improvement type projects that keep him wanting to get up. My husband is 50.

2004: My husband was diagnosed with MS ten years prior to being diagnosed with CADASIL. It was based on the results of an MRI. I am 49 yrs. old. I have had migraine headaches since I was 15 years old. My mother has had migraines for as long as I can remember. Her mother died due to multiple strokes. Her mother had her first stroke at the age of 40 and eventually died due to numerous strokes. I had my first stroke in 1996 at the age of 41. My younger sister passed away in 2001 at the age of 45 from a cerebral hemorrhage, bleeding stroke. My grandmother's brother had also died at the same age due to the same problem. I have not yet been diagnosed. In January, 2001, before my sister died, I became disoriented while driving and suffered a severe anxiety attack. I took a temporary leave of absence while many tests were done. There had never been any diagnosis given after the first stroke, I was just started on aspirin and Plavix every day. During these tests, it was found that I also have a hereditary blood disorder. This was blamed for my stroke in 1996 and the TIA's found from MRIs done in 2001. My therapist had found Billie's information online and gave it to me thinking this could be my problem. I am still out of work and suffer from many of the symptoms listed from her survey of 14 people diagnosed with CADASIL. Finally! I am making an appointment with my primary care physician AND neurologist and bringing along all of the information I downloaded from her website. I had mentioned this to my neurologist last visit. He asked me if I have passed out and I responded, "No", so he said I don't have it!!!! No skin biopsy offered, etc. Just no!! Well, I am a registered nurse, so docs telling me "No" doesn't stop me or deter me in any way, so NOW we will see what happens. Thanks for reading all of this! It is so nice to know that there are other people out there that are having the same problems AND are not crazy!!!!! I may be chronically depressed and many more things, but not crazy!

2005: Well done! The new website is wonderful, and I think you have done brilliantly to get the organization off the ground in such a short time span.

2005: I am a nurse working in the emergency department in a small rural hospital. I am also a family member of a family that suffers from CADASIL. In fact, all nine siblings of my mother's family have tested positive for CADASIL, along with many of my cousins. I am working on a presentation on CADASIL, and have a few questions. Why is thrombolytic therapy not recommended for the treatment or symptomatic control of

CADASIL? If a person with a known diagnosis of CADASIL presents in the ER with an acute neurological event, what would be the best course of action? Would treatment with TNK or TPA be helpful? Do you have any information from the nursing prospective that would be helpful?

2005 – Billie, our study is moving along, but we and the other sites need more patients. I am hoping you could do a second round of announcements to indicate that the study is still open for enrollment so we will be successful with the study.

2006 – Study is still going on. Placed on the website, e-mails went out and newsletters.
Over 255 patients were screened worldwide to participate in the study; approximately 168 patients have completed the study.

October 2007 – Steve is no several medicines to treat the symptoms. He is still on L-Arginine and takes it twice a day. His short term memory loss is decreasing. He is on Aricept a Namenda to help. We play with out grandchildren and enjoy each day as we can. I still am his case manager and I mourn each day of what I see in him. Steve gets tired very easy and has a nap in the afternoon. I work full time and worry about him.

Up to date information:
As of October 2007 – we have over 750 confirmed cases all over the world and over 215 doctors who have at least seen one patient who has CADASIL.

You can go to the website and look under confirmed cases to see where in the world are the confirmed cases.

If you are looking for a doctor look on the website for physicians/doctor locator.

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