

How my husband Steve is surviving his terminal illness called CADASIL.

This is how my husband describe the morning he woke up – waking immediately November 1994 early morning I was in the most excruciating mind spitting headache and I sprung out of bed. I finally although in the worst pain of my life of 38 years and in total darkness I slide out of bed and crawled to the bathroom finding the commode I vomited violently. Still in heavy pain and 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 left to rise up to standing position, to fumbled to switch on the bathroom light. Instantly in violent pain and still not knowing, I vomited into the sink and tried to crawl back into bed. **Steve was 38 years old.**

My husband had always been in great health and has had perfect attendance at work until 1994. I do remember my husbands 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.**

**Steve was 39 years old** In 1995 he had another major attack, which placed, in the hospital. This time the Catscan came back abnormal and a MRI was 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 this MRI come out this way, we will defiantly start to rule out Leukodystrophy, M.S., has migraines, maybe dementia or not sure and could be in his head. We will find out what this is, even if we have to research more, even if we have to use Lorenzo's oil." We came out of the doctors' office, stunned when the Neurologists words did not give us any hope.

Steve was functioning the best he could never complaining finding no answers or relief through morphine, Demerol or test after test. These test included spinal taps, blood drawn, etc. I did remember the words of the Neurologist and so my husband and I watched the movie "Lorenzo's Oil". Within a week I bought a computer and started research and 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 from the results of the MRI and CT scans. I got copies of my husband's files. Converted them to word and sent over 200 E-mails out with my husbands' history and asked for help. "Help is appreciated". Our first neurologist was so overwhelmed and amazed with the support and response he received from the e-mails, which were sent out.

I had great response from doctors, out there on the WWW. 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, what does it mean? Cerebral Autosomal Dominant Arteriopathy with Sub cortical Infarctions and Leukoencephalopathy, CADASIL is a diffuse disease of small arteries predominating in the brain. It starts during mid-adulthood and is characterized by recurrent ischemic events (transient or permanent), attacks of migraine with aura, severe mood disorders, and sub cortical dementia and, at MRI, a white spread Leukoencephalopathy, **TERMINAL ILLNESS AND AN INHERITARY 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 and my daughter's having a 50% chance of having CADASIL. There were about three items on the Internet concerning CADASIL only and the information related so much to my husband's symptoms. I hope it is not CADASIL maybe it is something else.

Dr. Hugo Moser, (known for Lorenzo's Oil) of Johns Hopkins University's 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'ed the MRI, CT Scans and all other documents to the Kennedy Krieger Institute to aide in his discovery. At the same time Professor Marie-Germaine Bousser was researching CADASIL in Paris, France she came across my e-mail. The Professor called me and requested copies of my husbands MRI and medical records that included my husband's family history. Though the Internet and fax machine I located medical records from my father-in-laws doctors going back to 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.

I had a force behind me as it took over 2 months to locate the medical supplies and dermatologist to perform this procedure and a radiologist to read the results. I did not stop calling everywhere to locate professionals who were willing to perform this test with instructions I had obtained from France.

Concurrently, Dr Moser had met with one of his colleagues from the Netherlands, having all the correct original documents and films from my husband's case and was confirming my husband with CADASIL.

**At the Age 40 1/2 years old** - July 1997, 2 days before the 2<sup>nd</sup> anniversary of my father-in-laws death the skin biopsy test confirmed CADASIL. **My Husband and his father who died in 1985 from a stroke** were both confirmed with 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 as the doctors could not say what he had, they said cancer first, may have dementia at 48 years old and finally 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 my husband has the same thing that her husband died of and Steve's sister and three other brothers might also have the defected gene?

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 on the Internet for more information on CADASIL, which is so hard to find. I have doctors and others e-mailing me wanting to know about CADASIL, as they are not aware of this disease. This is how I cope. When coming 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 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.

I love my family and scared of losing my husband and children, I want to protect everyone but when Steve was in the hospital in 1999. Our youngest daughter Noelle finally talked to me about Daddy. She asked 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 has 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% per chance of having it. She looked at me and said, like the game show "who wants to be a millionaire?" I explained how many people do become millionaires? **Then we talked about how Daddy has donated blood to create the first blood test for CADASIL to Dr. David Lynch in Penn University.** Hopefully they will have a test when our daughters get older to find out. She asked if there was going to be a cure. I explained that I hope to live for 70 years until they do. But how about daddy and a cure? It so hard to talk to her about this, I explained that if they don't have a cure for daddy at least 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 my get money, 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. 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 a director of stroke he turned and told me "I knew more about CADASIL then he does"! How can this be?

We live in not knowing when my husbands next TIA (small stroke) is or will he survive it. He has had at least one to two TIA's per year, which we know about. He did have a stroke in 2004, which left him weak on one side.

In December 2004 Steve's memory is going. He would say, did you feed the \_\_\_\_\_ you know what? Then point to the object. Saw the Director 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, New York University School of Medicine called me to see if anyone would be interested in the assisting with a study for CADASIL. He explained to me that the study would be for short-term memory loss and it is a medicine for Alzheimer's disease. I asked him if it was "Aricept", He said, how did you know. I explained that Steve was put on the medicine last month due to his memory. I said I would be happy to assist with getting participants. First ever Study for CADASIL Patients: The purpose of this study is to determine the safety, tolerability and 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 on 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 age of 25 and 70 years, and have cognitive impairment, are eligible to participate. The study will take place over 18- weeks.

Steve is functionally better than he was in the previously years due to all the 13 daily prevention medicines he is on, he does not live in pain. He still gets very tired, short-term memory loss, short temper (sometimes) and 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! We receive e-mails daily or phone calls for assistance or just to talk.

#### Inserts from journal

Steve had another mini stoke, October 18, 2003 - on Saturday morning he woke up light headed and within a hour told me he felt like he was drunk which included blurred vision and woozy. By the evening he was seeing double vision – which when he closed one eye would stop the double vision – He also felt queasy due to his double vision – We thought it might me a new medicine he was on – October 19, 2003 – Sunday Steve woke up still with double vision – he made a patch for one eye as he got tired of squinting – by the evening he was exhausted – I put a yahoo group e-mail seeing if any one else experienced double vision – This also encouraged me to contact his neurologist – His neurologist was not on duty but the other neurologist told us to go to the Emergency room. Steve didn't want to go as in previously we have been told that there is nothing no one can do for him, as CADASIL is so rare. I was insistent on going. We were in the ER at 10:00 p.m. and at 1:00 am he was admitted for observation. I carry the information on CADASIL in my car – and gave this to the doctors with a list of the 13 medicines he is on. When we got to the ER when we said possible stroke – they asked why we did not call the ambulance – He had a Cat Scan which showed nothing – all his other neuro signs looked good but his eyes started to go cross eyed more. They looked so different. The ER doctor told us he would contact our Neurologist in the morning and see what she thinks we should do.

Monday, October 20, 2003 – When I got to the hospital, Steve had not 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 and that she had talked with the ER doctor and told him to monitor his blood levels to do his sticky blood syndrome. 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 has had a mini stroke in the brain stem which causes the double vision and that he needs to patch the eye and every other day swap eyes with the patch for one month and if it does not get better then they will get glasses for him with a prism to assist his 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 – as we could off been still at the hospital unless I was active and was insisted with talking to his neurologist. It is so hard to here 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 and seeing Steve loose 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 only his eyes, which were affected with the mini stroke it could of, 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. I feel sometimes, Steve says things he does not mean as it's his illness but its so hard – even with the girls – 15 years old and 19 years old that they are losing their father also day by day. Thank you for reading this and I hope I can help others like other people have helped me. (October 2003)

Dr. Pastoures contacted me to let me when the locations of the studies kept opening. I had e-mail directly from other doctors asking for assistance with getting participants.

I was invited to attend a CADASIL conference 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, its over. I talked with Doctors, Professors,

Scientist, CADASIL patients, families of patients, etc. (Little o'le 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 have been e-mailing and assisted them in the beginning with CADASIL. One sufferer I e-mailed personally the week before the conference/symposium, without knowing I was going, and 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 and contacted me about seven years ago to find out what to do. It is a small world after all. During the evening meal, at the same table, I sat with the above families, a world expert Dr. Martin Dichgans from Germany. Dr. Martin Dichgans who got 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 how 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 gentleman who was in charge of the conference and had establish the CADASIL Trust in England.

He gave L-Arginine to me to take back to Steve to try it. I was sure it would not work as I have heard all the treatments other have tried. Steve starting 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.

A movie was just released call 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. Directed by [Alejandro Amenábar](#). Ramon girlfriend had CADASIL. Two people had signed the guest book to say they had watched the movie and was 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 was 200,530 hits.

Feb 2005 – 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 have 3 other children, two have CADASIL. The report I scanned and placed on the website under professional Health Pages so this will help doctors understand CADASIL.

March 2005 – over the spring break holiday – My sister and husband encouraged me to maybe start a foundation here in the USA. I went and bought the book “Non-Profit Organization for Dummies” and have not stopped.

March 2005 – 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 6, 2005 - A research Doctor from Germany e-mailed: 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 5, 2005 – Completed the Articles of Incorporation and filed with the State Texas. CADASIL Together We Have Hope Non-Profit Organization.

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 their 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 calls increased. And on May 10<sup>th</sup>, 2005 CADASIL Together We Have Hope became a non-profit organization ([www.CADASILfoundation.org](http://www.CADASILfoundation.org))

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 have 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<sup>th</sup>, 2005 – Received the Certificate from the State of Texas for Incorporation of the CADASIL Together We Have Hope Non-Profit Organization.

May 16, 2005 – received the first donation to CADASIL Together We Have Hope, Opened account.

Contacted testing sites for CADASIL all around the world (part of our mission) and ask if I could let other know about how to get tested. 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 as 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 for this disease. 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 – Newsletter were mailed out to over 300 addresses worldwide announcing the Organization, Study and letting other know about L-Arginine.

June 2005 – Completed the IRS Paperwork to file the 501 c 3 forms. See end of this letter to see the detailed form filed with the IRS on past, present and future activities for the organization.

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 – contact 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. I'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 and 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 coming 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<sup>th</sup> to July 24<sup>th</sup>. It was a wonderful experience. I have been enrolled with the ULF for over seven years now.

There were wonderful research, neurologists, and scientists, which presented at the conference. Some knew about CADASIL some did not. 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, Neurology and Pediatrics Director, 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 a number of U.S and International Investigators who may want to be part of this wonderful effort.

I met a couple whose daughter has MLD and had started a foundation. I was explained about one of the goals of the organization was to have a domain site. They volunteered to set up [www.cadsailfoundation.org](http://www.cadsailfoundation.org).

It is very important to know the mutation of your gene, which is affected with CADASIL. This will help generations in the future to be tested for CADASIL as a DNA blood test is only needed, as the doctors will only look for the defected mutation. There are 50 mutations in CADASIL, which is high. This is the reason why that it is so hard to find a treatment for CADASIL. The mutation can be inherited from one generation to the next. 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, 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 is he working on now and he said a rare disease called Fabry's. I could not believe this a friend of mines husband was diagnosed only 2 weeks before the conference. 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 last 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 and he said he was leaving tonight to go to that conference, as he was registrar. Before the presentation on CADASIL, you could say we had our first unofficial meeting in the USA for CADASIL with the person from Ohio, Nevada and a lady who just was tested as her brother had CADASIL and myself gathered about a table and talked for over 90 minutes about everything and the CADASIL Together We Have Hope Organization.

During the presentation on CADASIL, the presenting professor mentioned mine name several times saying that "Billie" has assisted with CADASIL or "Billie" can answer that question.

My 17 years old 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. At the time we know the mutation, we will locate a genetic counselor that knows about CADASIL and it will be my children's choice on weather to get tested now or later.

I have placed this information on the website: 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 is a 50% change of an inheriting disease like CADASIL.

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

On the foundation database there is over 264 confirmed cases by 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 - (<http://home.earthlink.net/~CADASIL101/tracking/>) There are at least 60 doctor who have seen at least one patient with CADASIL so if a person calls requesting a doctors name we can refer them to that doctor as we mail a information packet to. When attending the ULF conference I did notice I was already following the guidelines for HIPPA.

The response from the everyone was so supportive

July 31, 2005 – A doctor 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.

David

Posted on the website under Health Care Professionals – the PowerPoint presentation from the conference I attended. The doctor had forward me the presentation. I posted MRI, Skin biopsy reports also.

Registrar with Genetic Alliance as CADASIL was not recognized with organization.

Aug 3, 2005 – We need to have the American Stroke Association recognize CADASIL, sent the American Stroke Association a information package and they responded by this e-mail: Thank you for writing the American Stroke Association and sending information on CADASIL and 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; and 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, 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. And let's stay in touch.

Thursday, August 12, 2005 – I posted on the website the story at <http://home.earthlink.net/~CADASIL101/mag/> (please review)

I e-mailed out to all the people on the database and by Sunday, August 13 there has been 284 persons review this article.

August 13, 2005 – received this e-mail: Wow --- very interested 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 Disease 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. Received an invitation from N.O.R.D to attend this conference.

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". They have asked my husband to be a patient advocate.

Created chocolate bar wrapper on the computer to raise awareness all over the world.

August 21, 2005 – 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 the previously years due to all the 13 daily prevention medicines he is on, he does not live in pain. He still gets very tired, short-term memory loss, short temper (sometimes) and 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! We receive e-mails daily or phone calls for assistance or just to talk.

These are goals and what our foundation will be working towards and we are in the first stages of considering writing grant.

#### “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 Sclerosis 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.
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##### To educate and support doctors, patients and the public

- Educate doctors, radiologists, neurologist, and directors of strokes, and other 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.

##### Sample of CADASIL related e-mail 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 is what his father died from. And he also has a niece with the disease. He has two young children who could have CADASIL.

1998- a man who is 50 has been diagnosed with CADASIL. Has numbness in his feet. Is a perfectionist? Was a police officer for more than 10 years. He is located 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, and 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, his great-grandmother died of a stroke at 54 years old. He had a sister who also died at 26 years old.

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. 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 to 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 husbands 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 MRI and a skin biopsy that she insisted on as she has two young children, and is obviously concerned that they may pass it onto them as it is an Autosomal dominant condition. You are probably aware that there is little if any research being done in the UK but 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 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 there 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 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 5 and 8 and would like to be around awhile. Thanks

2000- Person 41 years old. Last year I was diagnosed with CADASIL. It has been a long journey for me and now I am having so many problems getting my medication for this. The doctors 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 might 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 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 leded 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 genetically 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. Physician well except for "mini-strokes" 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 out going 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 b/c 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 w/ 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. --HOLLY

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 guesses we are in 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 angeogram. 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 sub cortical white matter of the right anterior frontal lobe". In 1995 I started having two disturbing problems: a) memory problems and difficulty 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 and MRI angeogram, which has served as a baseline to compare the MRI and MR angeogram 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 not 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 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, whereas 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, but also to medical medias, neurologists, hospital teams, etc. In that purpose, we are preparing a Website (we will give you its address; it will be firstly in French, but we plan to translate it in English later), we will prepare a newsletter, get some contacts with medias, 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 [www.infobiogen.fr/services/orphanet](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 worse part of it and the fact that I forget a lot of things.

Believe me I write a lot of notes for myself and some times forget where I put them. The only thing the dr. 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 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 Minn 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 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 phone or write. He is like a child; he is only 34 yrs old. I hope this helps.

2002 My husband has had strokes since age 35. He is now 49. 8 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 anymore, 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 everyday 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 whom 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 anymore, 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 (heel 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 site. Thank you Billie for your site and for answering my 1st 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 catscan. The Catscan showed something and she was immediately taken for a MRI. Me - 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 too. 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 2-3 min 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 10 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 yrs. 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 Jan 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 MRI's 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 announcement to indicate that the study is still open for enrollment so we will be successful with the study.