1998-1999 Newsletter

1999 CADASIL Newsletters

IMPORTANT INFORMATION: MAKE AN ACTION PLAN . We did! We have to go pass two other hospitals, but we will be very demanding if Steve shows any signs or symptoms of CADASIL.

I received this information from Eileen Knox - I will not name the hospital but wanted everyone to know. If you do go to the doctors take information on CADASIL with you! It is not my intention to scare anyone, but this is for you information. Eileen Dad had a spell. He thought he was having a stroke so we took him to a hospital. They sent him home said it was an inner ear infection. By the time they got him home, he started having another stroke, so they rushed him to another hospital. The doctors knew just by looking at him, he was having one. Now he is blind in one eye and his face is drawn. He is still in the hospital. We expect him to be out in a couple days.

We take the newsletter with us along with our reports from the doctor. They act so dumb founded. It really worries me because too many mistakes can cause things like what happen to my dad. still have a hard time with the doctors when I go into the ER, with my health problems. They act like I am too young to have strokes and such. Like I'm lying. Well if they seen my MRI they would think differently.

THANKS EILEEN FOR LETTING ME SHARE THIS. IT HELPED US DECIDE WHAT TYPE OF PLAN TO MAKE JUST IN CASE.

Since the last Newsletter: There is an online email news information, talk at http://www.onelist.com for family, friends, etc. who in one way or another are effected by CADASIL.

This on-line is also for doctors who have shown an interest in the disease. This was created by Doreen Natte ter-dor-nat@webtv.net or dornat1@yahoo.com who has been diagnosed in 1995 although she is in her 13th year with having it, Her mother has been in a nursing home for the past 11 years after having severe strokes caused CADASIL. The purpose of this list is for people to share their feelings and thoughts and as an outlet for us that have CADASIL or have family or friends with it. Sso many times there is no one who knows what we're going through, and this list is a place to go that is always there to talk night or day. This is not a chat room. This is strictly an email group.

HOW TO SIGN UP: When you post a message to the group it goes to all the members of this group, or you can choose to email individually. To sign up, simply go to: http://www.onelist.com/subscribe/CADASIL If you are not already a member of Onlist,

you will need to register. On the left side you will see a menu bar. Near the top you will see "new member". You will then be taken to another page where you will enter your email address and choose a password for yourself that you will remember and follow the remaining instructions to become a member. After receiving verification that you are now a member, go back to www.onelist.com/subscribe/CADASIL and enter your email address and password. Go down to the bottom of the page and click on "subscribe to this community". You will be asked to sign up for "normal" version or "digest" version on the next page. (Normal version means you will be e-mailed each message as it is posted and Digest version collects all messages and sends them to you in one large e-mail which is normally sent once a day) For those of you who are already members, follow from *** on down. After you sign up, you will be sent an e-mail verification that you are now a member.

This was created on 8/25/99. You don't have to post until you are comfortable doing so or you can publish your story in the next newsletters **cadasil@earthlink.net** Some people enjoy this and other want to "lurker" and read what others post. That's okay, too. You are under no obligation to join as with anything doing with CADASIL. Myself it is a good way to communicate with others, but can be very scary when you know what might happen in the future and sometimes I even get nervous about looking at this type of e-mail.

United Leukodystrophy Foundation (ULF) If you would like more information on ULF, you can contact them at 1-800-729-5483 in the United States or International 1-815-895-3211. They send newsletters out every quarter and deal with all the Leukodystrophy diseases. The leukodystrophies are genetically determined progressive disorders that affect the brain, spinal cord and peripheral nerves. The term Leukodystrophy derives from the Greek words "lueko" meaning white and referring to the white matter of the nervous system. "Dystrophy" means imperfect growth or development. If you know more, please let me know. Usually when CADASIL patients are first evaluated the doctors rule out M/S or another dystrophy as CADASIL is still a new disease.

Others links: If you want to link to others like us or want to be on the list for the support group contact, Billie. Please e-mail or mail me your e-mail or address.

Billie and Steve Duncan-Smith CADASIL support group newsletter
3605 Monument Drive, Round Rock, TX 78681
512-255-0209 home

You can contact other families who are also going through this awful condition.
REMEMBER YOU ARE NOT ALONE look at the attached list or go to
www.home.earthlink.net/~cadasil/sup.htm

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.

Steve's progress:

Steve is slowly changing, but I can see it. The attacks hit him, with dizziness, throwing up, and migraines. He never complains and says oh by the way, I had a spike today. When he gets bad during the day, he take Ala-Sseltzer to reduce the pain. He says there is no point in going to the doctors because they cannot help him. He does not complain but I know when he is in pain. He gets upset so easily that everyone gets frustrated.

Steve does not complain but he does wake up with a headache each day and it 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 nightmare and everything will be normal. He has lost more weight and seems to has lost hair from his head. He seems so weak sometimes and with his l weight reduction, his skin feels so different. He always seems cheerful. The thing that I have noticed more is that he gives in to our daughters so easily. I think he is scared that he won't be able to see them grow up. His own doctor told me he does not know what else he can do for Steve. (1999)

CADASIL EMAIL'S:
We'will call my husband GEM, because he is. GEM held a managerial position at a fairly large corporation, where we met. In 1990 we were married. Both our second marriages. We were in heaven. Then, out of the blue, the nightmare began. Suddenly, one year after we were married, in August of 1991, GEM (age 46) became very ill with a severe headache and vomiting. I took him to emergency. Thinking it was a severe migraine the doctor gave him strong painkillers. The next day he was totally confused. He didn't't know my name or his own name. I took him back to emergency. He was admitted to the hospital in ICU where he stayed for five days with every test imaginable being done, to no avail. On the fifth night he suffered a seizure and the next day he woke up and said, "what day is it?" The doctors attributed the cause as being a virus that somehow entered the brain. GEM had suffered from migraines since he was 16, but otherwise, he was just a big healthy sweetheart. During his six-week recovery, on a follow-up to his neurologist, he mentioned that he had suffered from migraines, so his doctor started him on Verapamil. He still suffered from migraines, but not as frequently or severe. His migraines always seemed to come on <u>after</u> the stress factor. He had a 98% recovery and was back to work and pretty much as good as new.

Then 8 years later, in August 1998, he became very confused at work and was sent home. We tried taking some time off, but he became more and more confused. So back to the doctor. GEM was treated with 5 consecutive days of prednisone infusion, which after 3 days showed marked improvement (a miracle medicine). We were informed that the "footprints" on his MRI from 8 years before had increased and that it was possibly an "atypical" MS. Not wanting to accept this, we requested another opinion. Our doctor referred us to the MS specialists at UCSF, Mt. Zion (San Francisco).

The specialists at Mt. Zion, agreed that it was not't a typical MS, and thought it might be MELAS, a mitochondrial myopathy. So a muscle biopsy was performed, but came back

negative. During the pre-op for the biopsy it was discovered that GEM had an a trial fibrillation, so he was started on blood thinners and other medicines for his heart.

Then in May of 99, he started getting a headache that would not't quit and became confused again. We rushed to his doctor, both crying like babies. We were like two scared little children running to their dad for help. The reason was that it had taken almost a year and he was finally improving, but now another set back. I guess we thought that if this only occurred every 8 years or so, we'd be OK for a while. This time he was promptly treated with another 5 days of prednisone infusion and bounced back very well. The MRI showed only a minor increase.

Now to the part where the CADASIL comes in. A few months ago, GEM's older brother, by 4 years, who had been in perfect health, suffered from two mini-strokes. Interestingly, his brother's MRI showed something unexplainable. His neurologist, out of St. Louis, suspected CADASIL. A skin biopsy was performed and the diagnosis was confirmed.

GEM had four siblings, three brothers and a sister that died at age 11 from a brain tumor. His younger brother, by four years, has been epileptic since the age of 28, but is otherwise healthy. Their mother, who suffered from strokes, passed away at the age of 74. His father still lives and is 91 and one-half.

GEM has three beautiful daughters, ages 28, 24 and 18. The two oldest daughters were recently married. The 28 year old suffers from occasional migraines, the 24 year old became epileptic at 17 and the 18 year old suffered some unexplained illness last year, with an abnormal MRI. Both of his brothers each have two sons, in their twenties, who are healthy.

GEM has become permanently disabled. He does not't work anymore. He can walk around, but tires easily. He never gets headaches anymore. His speech is slurry, sometimes worse than others. My right is his left, and vice versa, so he does not't drive anymore. He says he does not't feel bad, just tired. His personality has definitely changed. He keeps busy in between naps; he plays games on the computer and watches videos, which the sad ones always make him cry. He loves to watch game shows. His long- term memory is amazing. He's been busy gathering info for a genealogy study.

From reading all of the "letters", it seems that we're all at different stages of this disease. It also seems as if it must be "hit or miss" genetically and there are many varying degrees of symptoms. I agree that the general public needs to be informed as they are about MS, Alzheimer's, AIDS, etc. I strongly feel that the children should be informed so that they can at least have a choice whether or not to take the chance of passing on this dreaded disease.

Well, now it's time for us to take the test. It was so frustrating not knowing what it was, but honestly, it's not any better knowing what it is. But to finally know that we are not't alone *is comforting*. Caringly, G&L

Story from U.K. I think that the majority of doctors and consultants probably do not know any more about Cadasil than we do and can only offer limited treatment which will help with circulatory problems and pain relief until more is known about treating the cause.

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's and Steve's site)

About how long it takes to get results of DNA tests back - I asked my father and he said that 2 months was about the minimum time (my mother and sisters tests took nearly 6 months, but that was 2 years ago). Now that they know what they are looking for it should be quicker. Try not be scared - I know its hard but if you can stay positive you can help your husband though the bad times - I thought my mother was going to die last year after she had a bad stroke but she got through it with the love, support and encouragement from all of us.

I know what you we are going even though I do not have Cadasil. I always hope that when I call my mother and ask how she is that by some miracle she will say she feels well - but sadly she always says how 'drained' she feels or how bad her head has been. Luckily she somehow manages to stay positive and is determined to fight as best she can against the affects of Cadasil. Knowing about this web site has renewed her hope that a cure will be found - I'm positive that it will and that the more the word is spread and the more the awareness is raised, the sooner the cure will be found. Its good to see new members signing on - lets hope we can reach out to the people who can make a difference. Mark son of Richard Pritchard.

Story from France - Hello ! My name is Chantal NEAU. I live nearby Paris. I am a cousin of Nicole JOLLET and we belong to the family in which the gene of CADASIL was discovered. I have many relatives who are hit by the disease, but my father hasn't got the gene and I am therefore lucky to have no risk of getting it. But I'm sure it influenced me a lot when I was a child to see my grandmother who was paralyzed. CADASIL create sufferings also to people living with persons who are ill... My cousin forwarded to me an e-mail from you. I have subscribed to a Cybercafe (my e-mail address is chantal_neau@plaisanceweb.com) and I'll try to communicate with you by Internet. If you think this mail is interesting for other people belonging to the CADASIL Onelist, you can of course forward it.

Now, there are 80 families in France who have been identified with the same genetic problem. The medical research is quite active here, and there is also a German team working on it. We have created an association in order to create a link between the families, to collaborate with researchers, and make the disease better known in the medical society. We will create some documentation, probably in French, but we plan also elaborating a web site, and I will try to help in translating it. We will be happy to exchange some information with other people abroad, using for instance your list. Many

people feel alone, need some help, understanding, knowledge, and reasons for hope.I am a participant in this new association (ACF France for Family Cerebral Arteriolopathies) and I will do my best to help creating a big worldwide chain of communication. French researchers have faith that the disease will be treated in the future, and we have to follow their work, collaborate with them, and communicate their results to all persons concerned by this disease! Yours sincerely, Chantal

Another Story - It is a long time since I've been in contact with you, Billie, but I think I'd better update you on how things are going down this end. Since we last communicated, I have had at least one more stroke which has left me all but unable to speak at all. I also seem to have developed a type of dementia which has affected my memory rather badly, so that I cannot recall events that may have taken place as recently as 5 minutes ago; the same dementia is also responsible for my confusion as regards location: my wife has to go everywhere with me for fear I won't be able to find my way back home again. As regards driving it is very difficult at the best of times. One of these days I suspect I will kill myself, so that I don't have to worry about death from natural causes at the hands of Cadasil.

A facet of this dementia is my almost total confusion in normal everyday situations: things that I used to organize without so much as a second thought have become major logistical exercises: for instance, making a cup of tea is no longer very easy because it takes working out what order everything is done in and once I have worked this out, I still have the problem of finding out which cupboard the materials are kept in! Which reminds me,writing this is very difficult because I constantly have to remind myself of what order the words come in - reminding me how difficult yet so effortless is the business of language, at any rate for the healthy mind. I wonder how long it will be before I lose my mind completely. At least I am still sufficiently compos mentis to keep one train of thought running long enough to sustain the ability to compose a reasonably coherent sentence! I wonder what happens when even this ability deserts me - I guess I will not be able to communicate at all. What a depressing thought! I entitled this note "genetic testing", so I guess I'd better get down to it.

A scheme has just got under way in this country United Kingdom whereby the Cadasil problem has just begun to receive proper medical attention and attracted serious interest for the first time.I was diagnosed with Cadasil and told I was suffering from a genetic mutation in 'Exon 4, Notch 3', which helps to explain my condition and, worryingly, is confirmed as autosomal dominant. My children are therefore at risk of inheriting the illness. This all sounds rather technical, but what it amounts to is a definite diagnosis of my illness. It is not a great consolation to know that I am not barking mad- that there is a perfectly good reason for my apparent strange behaviour- but it is rather difficult to appreciate it at times. Apart from my neurological difficulties, I'm also suffering from a numb right leg, which I find irritating rather than debilitating. This came on about 2 months' ago and occasionally causes me pain insofar as it often appears that I have got a band of rigid muscle underneath my right foot; fortunately, this is the limit to my physical discomfort. So, as you can see, my condition has deteriorated a little bit, Billie, since our last communication. I do hope that you and Steve are both in good health and certainly no

worse than you last reported. I also wish to take the liberty of getting in touch with one or two of the other self-confessed sufferers from Cadasil too. Maybe we will be able to compare notes on our respective illnesses, and perhaps be of some moral support to one-another. Regards and best wishes, A ex-educator in England.

Copy this newsletter out and give or mail it to your doctors or anyone who is in the medical professionals so hopefully one day their will be a cure.

Please remember this newsletter is to help others. I do not want to mislead anyone. I am looking for HOPE, link to others with CADASIL, find out as much about this disease as possible and hopefully a cure one day. I am not in the medical field or claim to be a professional on CADASIL.

This newsletter goes out to people in America, England, Scotland, Sweden, France, Chile and Australia. If you know of anyone who is interested in this newsletter, please photo copy it, and pass it on. Also, contact me so we can add their name on the email lists or mailing address..

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 husbands illness. When produce these newsletters I sit at the computer and sometimes cry but it does seem to help the stress.

If anyone can help to clarify anything dealing with CADASIL, please let me know or provide any advise or help, please let me know.

Thank you for reading this newsletter.

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CADASIL SUPPORT

GROUP NEWSLETTER

The unofficial CADASIL newsletter Issue IV June 1999

CADASIL - Cerebral autosomal dominant arteriopathy with sub cortical infarctions and leukoencephalopathy Recently identified, 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, sub cortical dementia and, at MRI, a white spread leukoencephalopathy.

Since the last Newsletter:

I have had trouble trying to get the new web site location at Http: //home.earthlink.net/~cadasil/, responding to the web searchers. A few of the web searches will bring it up under the name search "CADASIL".

I have had wonderful response from the web site support group and please take the time to see if you have anyone on the support group close to you. Also, let us know if you have contacted others like us.

If you do not have the access to the Internet, please let me know and I will mail you the newsletter. Otherwise you can find this newsletter on the web site. http: //home.earthlink.net/~cadasil/ (you can find the ~ symbol on the left side of the number one key on your keyboard.

I have already published all the newsletters on the web site now and if you would like a print out of them go to each one and hit print. Once again if you are not available to the internet, please let me know. Remember you are not alone with this awful condition.

United Leukodystrophy Foundation (ULF)

If you would like more information on ULF, you can contact them at 1-800-729-5483 in the United States or International 1-815-895-3211. They send newsletters out every quarter and deal with all the Leukodystrophy diseases. The leukodystrophies are genetically determined progressive disorders that affect the brain, spinal cord and peripheral nerves. The term Leukodystrophy derives from the Greek words "lueko" meaning white and referring to the white matter of the nervous system and "dystrophy" means imperfect growth or development. If you know more, please let me know. Usually when CADASIL patients are first evaluated usually the doctors rule out M/S or another dystrophy as CADASIL is still a new disease.

Research

I have not heard about any more research since the last three newsletters. If you know anything please let me know and I will publish it on the web site immediately. The last letter I had was stating that soon drug trials may happen. I have no idea when.

Others links: If you want to link to others like us or want to be on the list for the support group contact Billie. Please e-mail or mail me your e-mail or address.

Billie and Steve Duncan-Smith
CADASIL support group newsletter
3605 Monument Drive, Round Rock, TX 78681
512-255-0209 home / 512-428-2901 work

Ralph and Lorraine Hoyle, P O Box 404, Carmel, CA 93921 408-624-1911

Kurt Shonka 845 Mandalay Road, Jacksonville, Florida 32216

William Freeman, 12, Hatten Ave, Rice Lake, Wisconsin 54868

Mr & Mrs Richard G Pritchard, 99 Christine Avenue, Rushwick, Worchester, England WR2 5ST

For others names and info go to www.home.earthlink.net/~cadasil/sup.htm

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.

These are medicines which others who have CADASIL are on: Remember there is no cure for CADASIL as of yet but Together we can have HOPE!
one tablet 225 mg alpha lipotic acid,
one tablet 325 mg Bayer Aspin,
one tablet 1000 mg Vitamin E
Papaverine 15 mg one tablet twice a day, Depakote 250 mg one tablet at 11:00 a.m. and 9:00 p.m Please check with your doctor before you change medicines, this is what others have tried or use.

These are medicines (BW) another patient is on:
3% Hypericin 300 mg of St.John's Wort 1 tablet a day. It really has seemed to help even out his moods and he doesn't seem to swing so far. 2000mg Vitamin E, 325 mg buffered aspirin.

Please check with your doctor before you change medicines, this is what others have tried or use.

Other patient daily medicines: (u)
Volmax 8 mg tablet Cardizem CD 300 mg capsule Zocor 10 mg Baby Aspirin, Clonazepam 0.5 mg, Vitamin E ,Trazodone HCI 50 mg ,Folic Acid 1 mg ,Fibercon 625 mg, Dexamethasone 4 mg Senokot-S Tab ,Prevacid 30 mg

It is not proven but when Steve gets a bad headache attack he takes an Alka-seltzer. This reduces the pain a little. We read this in a magazine.

Steve progress

The last months has been more of a nightmare, he has had several attacks again. The attacks hit him, with dizziness, throwing up, migraines. The attacks can last up to 5 days. Some mornings when he gets out of bed, that's it. He never complains but its so hard to see your husband in pain. When he gets bad during the day, he takes Alka-seltzer to reduce the pain. If he wants to be knocked out , so he says he takes Alka-seltzer PM. It seems to help him. He says there is no point going to the doctors because they cannot

help him. He does not complain but I know when he is in pain. We play a game called 1 to 10. 10 being the least pain. Normally he says 7 to 8.

Steves temper has shortened. He gets upset so easily that everyone get frustrated. Our 11 years old is so short tempered. She says, but daddy is also. How do you explain that daddy has a illness which he cannot control his temper and some day you might have it also?

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 nightmare and everything will be normal. We just had our 20 year anniversary. I love him and do not want to lose him.

This story: Our family were totally clueless on what was happening to my dad. They had found white matter on his brain. His family doctor had recommend him to see a doctor in Minnesota. My mother had talked to the doctor and he wanted all of us to come out and be tested. We were all having migraines, strokes and weakness in our legs. My brother, father and sister went to Mayo Clinic for two weeks of testing. They got the results back. My dad and brother were diagnosed with CADASIL. As for sister because of her age 14 they would not reveal the test results because they said she was to young to deal with this. My other brother and I went to Columbia, Mo for testing. We found out our results in July 1998. I was diagnosed with CADASIL. My brother's MRI didn't show any abnormalities. I know he has it because he has the same problems as all of us. He is planning a trip to Mayo Clinic for further testing such as skin biopsy. This disease has robbed us of our father. It is a nightmare we can never wake up from. I'm 26 years old and have over 26 places on my brain of the white matter.. My brother is 31 years old and has over 37 places on his brain. My dad has really changed over the years. His mood swings are worse. He has trouble seeing and has no strength. My father has forgotten how to do things he has always done. I pray each night that they will find a cure fast. We are not the only ones in our family that has been diagnosed. My cousin Ms. Bryd from Arkansas was diagnosed with CADASIL by the same doctor. There are about 25 or 30 in our family on my father's side of the family that has CADASIL. This has really been so hard on us. My mother is the one who is really hit hard by this. It is a struggle everyday for her. I know my dad can't help what he does or says. Its like someone has taken over his body. My mother is in counseling right now. She is trying so hard to be strong for all of us. My father seems to talk a lot about death since he found out he has CADASIL. He has given up hope. I'm bound and determined to take care of myself to hope that I never get his shape. I go to bed every night wondering if I will be able to take care of my daughter and if she has this. I was approved last year for Disability , lucky it took just three months. Our neurologist, has put us on Depakote , Paxil, Ativan, and Neurontin. We would love to hear from anyone. Senora Stockton, 603 East 10th Rear, Portageville Mo 63873 phone # (573)379-5499 e-mail stockton@sheltonbbs.com

Follow up story: Right after Christmas we went for 2 weeks to Ann Wigmore's Institute in Puerto Rico. It is a holistic type health institute. What have we got to loose, nothing else is really working. It was a wonderful experience. My husbands blood pressure

dropped dramatically while we were there and by the second week he was lifting up his feet instead of shuffling them, he was climbing steps (which we avoided) his color was so much better and he was speaking better. Our children noticed the difference as soon as we got home and, of course, everyone else did too. My husband is a little reluctant to chalk it completely up to the institute because he still feels like his head is swimming around inside. That feeling never left him. Since we have been back he has been taking short walks through the house without his walker. The institute's focus is on raw food(uncooked) and the enzymes are not killed. All food is cut up small,blended, grinder or very chewed. This is suppose to help you body with digestion, which is a lot of work, and allows your body to concentrate on killing diseases and healing. It makes sense. They also work on your well being and self esteem. There were many wonderful testimonials of people who had incurable diseases (by medical standards) and were living proof that this works. We saw them. Cadasil is a little different say than cancer, but one of things that they work on is vascular problems. Nothing will make me believe it didn't help. He has been exercising at the gym at least 5 days a week for about a year. Mostly working on upper body strength but does the bicycle and tread mill (the kind you can hold onto the bars with). We are both convinced that the exercise is helping to keep him well. He has had no more episodes since July 1998. One night he got up and could not't walk, but he was fine in the morning so it must of been that he was in such a deep sleep.from the second newsletter:

A note from a Doctor:
I was the doctor who introduced the ULF to the concept of CADASIL last July with my presentation at their annual scientific meeting in Sycamore, IL. I am aware of 5 families and will send each a copy of your newsletter. Hopefully, they will then contact you. My department has a Neurogenetics Division that provides care to patients with inherited disorders of the nervous system including CADASIL. To confirm the diagnosis, we use skin biopsy that is examined by a superb pathologist at the New England Medical Center, an individual especially experienced in the study of skin ultra structure. We are also banking DNA from affected individuals and are planning to do studies on the NOTCH3 gene. You may feel free to suggest us to patients in the New York-New Jersey-New England region. Sincerely, Edwin H. Kolodny, M.D. Bernard A. and Charlotte Marden Professor and Chairman, Department of Neurology New York University School of Medicine Tel.: 212-263-6347 Fax: 212-263-8228

SURVEY RESULTS SO FAR:
Does this sound like any of the symptoms you might have or you know someone with CADASIL might have or had:

1. What type of symptoms did you have before you found out you had CADASIL?

1. **Headache on one side, numbness in left side** (dc)
2. **Stroke at the age of 35, no cause, dizzy spells, bad headaches, numbness, tingling in legs, hands, mini strokes. (ND)**
3. **Mini Strokes & Seizures (WF)**
4. **Migraines, dizzy, sickness**

2. How old were you when you symptoms first started?

1. **35 years old**
2. **35 years old**
3. **48 years old**
4. **38 years old**

3. What other suspected diagnoses did you receive, before you were diagnosed with CADASIL?

1. **Stroke in 97, MI in 95**
2. **Depression, getting along with others,** osteo arthritis
3. **Stroke**
4. **Migraines, doctors said m/s or** lad

4. How old were you when you were finally diagnosed with CADASIL?

1. **37 years old**
2. **45 years old**
3. **49 years old**
4. **39 years old**

5. Are you working full or part time?

1. **neither**
2. **no, permanently disabled receive social sec benefits.**
3. **no, severely disabled, social sec benefits.**
4. **full time**

6. How old are you now?

1. **37 years old**
2. **47 years old**
3. **50 years old**
4. **42 years old**

7. Are you a perfectionist or would you say you want things a certain way?

1. **yes**
2. **I am definitely a perfectionist**
3. **no**
4. **yes**, **wife says definitely**

8. Do you suffer from allergies? If yes, what type?

1. **yes - penicillin & food**

2. **yes - penicillin & cigarette smoke**
3. **no**
4. **yes - environmental**

9. Do you suffer from sinus problems? If yes, what type?

1. **yes -does not drain too well**
2. **yes - sinus headaches**
3. **no**
4. **yes - drainage**

10. Do you have back pain? If yes, where?

1. **yes, lower back**
2. **yes, lower back**
3. **no**
4. **yes, lower back**

11. What medicines are you presently on?

1. **Ticlid,heart med., vit e, b2, eltroxin (thyroid)**
2. **1 aspirin, folic acid (for high homocysteine), Motrin, Verapamil(for heart)**
3. **Vitamin 3, Ticlad, Baby Asprin**
4. **225 mg alpha lipotic acid, 325 mg Bayer Aspin 1000 mg Vitamin E**, **Papaverine 150 mg & Depakote 250 mg**

12. Do you get frustrated easily?

1. **yes**
2. **YES! Easily**
3. **No**
4. **defiantly so**

The above survey will hopefully help others know the symptoms of CADASIL and also help the medical professional understand CADASIL better.

Print this newsletter out and give or mail it to your doctors as the more medical professionals know about CADASIL then hopefully one day their will be a cure.

Please remember this newsletter is to help others. I do not want to mislead anyone. I am looking for hope, link to others with CADASIL, find out as much about this disease as possible and hopefully a cure on day. I m not in the medical field or claim to be a professional on CADASIL.

This newsletter goes out to over 40 people from the American, England, Scotland, Sweden, France, Chile and Australia. If you know of anyone who is interested in this

newsletter, please photo copy it, and pass it on. Also, contact me so we can add their name on the email lists.

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 husbands illness. When produce these newsletters I sit at the computer and sometimes cry but it does seem to help the stress.

If anyone can help to clarify any of the questions which are still unanswered, please let me know or provide any advise or help.

Thank you for reading this newsletter.

DON'T FORGET TO TAKE THE SURVEY at
http://www.home.earthlink.net/~cadasil/sur.htm

Contact Billie at CADASIL101@HOTMAIL.COM

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CADASIL SUPPORT

GROUP NEWSLETTER

The unofficial CADASIL newsletter Issue IV February 28, 1999

CADASIL - Cerebral autosomal dominant arteriopathy with sub cortical infarctions and leukoencephalopathy Recently identified, 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, sub cortical dementia and, at MRI, a white spread leukoencephalopathy.

Due to changes with my computer I had to locate another Internet provider so the home page has changed to http: //home.earthlink.net/~cadasil/ You can find this ~ symbol on your keyboard by the left side of the #1 key and you will need to also hit your shift key. The style of the web page has changed.

I now have the ability to publish the newsletters to the web site so you can now print them and pass them out to your family, doctors and friends. If you do not have the access to the Internet, I do not mind mailing the newsletter to you. Just please let me know. I have already published last February newsletter and plan to publish the other two soon.

I want to thank everyone for the best wishes concerning my wrists. I have had one surgery and will be having another surgery next week. Please give me a couple of weeks before I can respond to your e-mails. Remember you are not alone with this awful condition.

United Leukodystrophy Foundation (ULF)

If you would like more information on ULF, you can contact them at 1-800-729-5483 in the United States or International 1-815-895-3211. They send newsletters out every quarter and deal with all the Leukodystrophy diseases. The leukodystrophies are genetically determined progressive disorders that affect the brain, spinal cord and peripheral nerves. The term Leukodystrophy derives from the Greek words "lueko" meaning white and referring to the white matter of the nervous system and "dystrophy" means imperfect growth or development. If you know more, please let me know. Usually when CADASIL patients are first evaluated usually the doctors rule out M/S or another dystrophy as CADASIL is still a new disease.

This web page is how this web site started and how others cope with CADASIL in their own words:

The stories are published in the newsletters.

If you would like to tell your story, please e-mail me. cadasil@earthlink.net I will leave your name/s off the story if you wish since I want to respect everyone's privacy.

Please remember this web is to help others. I do not want to mislead anyone. I am looking for hope, link to others with CADASIL, find out as much about this disease as possible and hopefully a cure one day. I am not in the medical field or claim to be a professional on CADASIL.

This is my story about CADASIL, how I found out my husband has CADASIL and why I established the Web pages: I have moved this story to the home page, please click on home to read about Steve and his symptoms, doctors and how I cope. home.html

STORY #2

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 Radio phone 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. Mr R G Pritchard, Worcester ENGLAND

STORY #3

Thanks for sharing and all the hard work that you have done on you and your husband's behalf. My name is K and I live in USA. I first heard about Leukodystrophy in 1989 when my 59 year old mother was diagnosed with an unspecified form. This was before the Internet was accessible to everyone so being in health care I went to the medical library of a local hospital. I didn't find much but as a son I got plenty scared. My mother moved when my dad retired and hooked up with a neurologist in our state. He was interested in her case and followed her for the next nine years until her death in January of this year, 1998 at the age of 68. My dad made arrangements with the doctor to have my mother's brain autopsy and sent to a university. Only after several months did they determine that she had what is now described as CADASIL. For myself this would only be a sad and sorrowful story of the loss of my mother. However, in the spring of 1996 while going through a most stressful business merger I began to experience some localized left side parasthesia in addition I began to experience some visual distortion consistent with migraine episodes. Also, during this time I attempted to give blood at a church drive and was denied because my blood pressure was too high. The parasthesia broadened and became bilateral at which time my family practice Dr. referred me for an MRI. They were looking to rule out MS also. What they discovered was large areas of abnormal electrical activity but nothing definitive. I was further referred to a neurologist for a complete neurological exam and work-up. I was given a lumbar puncture and several tests which I cannot remember the names. All of these tests were negative. I had my father deliver the MRI's which my mother had taken in '89 and the two were compared. That was when the Drs. became suspicious that they were dealing with a disease similar to my mothers. I was referred to the Mayo Clinic in Jacksonville where I live. They did an exam and found me neurological intact but compared the MRI and determined the similarity also. She took lots of blood and had it tested for all kinds of calamities all coming up negative. The most recent

finding are pointing toward having a blood sample forwarded to be tested for the genetic marker to confirm the CADASIL. In addition, my mother's mother suffered severe dementia and died in a nursing home at the age of 78. Currently my mother's younger brother age 65 began suffering specious neuro symptoms consistent with my mothers. The same doctor also told me that he is aware of a woman in Orlando who because of a recent car accident had an MRI and they found a similar pattern of electrical activity. There is also some good news to my story. In 1996 in the early summer I began to fight back against the stresses in my life. I took a stress self-regulation program based on John Kabbat-Zinn's program at the university of Mass. medical school focusing on Mindfulness and Meditation. I dropped several pounds on a high protein diet and began a slow and painful harnessing of a Trojan work schedule. Today, I have a business partner and we extracted our practice from the merger to form our own corp. I have reduced my work schedule significantly and increased my recreation. Currently I am symptom free. I still am. I have two teenage children and a wonderful wife and I want to hope for the best but prepare for the practical. I hope this short story is helpful to someone. My name is Kurt Shonka. My address is 845 Mandalay Road, Jacksonville, Florida 32216. My phone # is H 904-724-7321 My E-Mail address is jaguar@tu.infi.net. I look forward to communicating with you all as I learn more about the condition and my own. Best wishes, KURT

STORY #4

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 Night-line 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 thought 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. L.W-

STORY #5

Today in searching the Internet for information on CADASIL I ran into your web page. God Bless you for now I don't feel alone with this awful disease. In April of 96 while driving my mother to the clinic my wife had a seizure and after being in the hospital and doing tests a MRI showed that she had suffered a stroke. Her family background showed that her grandmother on her mothers side also had strokes

early in life, My wife was only 48. In addition my wife mother had just recently 4 or 5 years ago, had started having mini strokes, she is 70. My wife Diane since that time has had many mini-strokes that have taken a toll on her mental abilities and Doctors were unable to find out what was causing them. 1 year ago her Doctor sent her to a hospital here in W that was highly respected and after consulting with Doctors at this hospital they said unless we could find out what was causing them, they would continue. Sept 25, 1997 D underwent brain surgery in attempts to find out the problems. Its was after the test came back from the Mayo clinic and our hospital that the Doctor said she has CADASIL, no treatment, no cure, no future, and most of all no information from which to find out anything about this disease. I have to tell you that I was crying after finding your web site and reading about your husband as I know how you feel and what you are going through. D, thank god has no headaches, has undergone a personality change, suffers greatly from typical stroke problems, but most of all has severe dementia. I have to dress her daily do the cooking and housekeeping etc and care for her like a nourishing home would a person with Alzheimer's. Thank god D has no pain and does not know what is happening to her, except she tells me that she is not getting better. She seems to bounce back in between strokes ,but never back to where she was before the last one. I wonder when the next one will happen and how bad it will effect her, as she has not much of a mind left. Thank god she is still company to me and we can do some things together, even though I have to tell her when to eat, what, when to go to the bathroom (if I am not to late) and her close friends still stop and see her even if she is not much company to them. D is being treated with a drug that helps the small blood vessels in the brain stay open to prevent clogging (ticlid) also 1 baby aspirin daily. Does this help? I doubt it but its something as the doctors don't seem to know anything about it and they is no apparent cure, I don't know how much longer I am going to have my wife as I am a 58 year old retired Police Officer with a 31 year old daughter who has 2 children and a son 28 who has a 2 month old daughter. Doctors say this disease is passed on from family to families and I worry daily about their future. If you have any additional information on this disease I would like to have it. Thank you so much for thinking of others who are going through the same hell as you. At least we are not alone. Again thank you for letting us share in our grief together. After reading all of the material of the web site, I am very much interested receiving your newsletter, which after reading I intent to pass on to my wife doctors. As you probably know, they know little about this disease and seem too busy to research it further, telling me that its not even in the medical books, and they just recently have heard about it. again, thank for your wonderful work as I will continue to find out all I can about this dreaded disease.

STORY #6

My wife has CADASIL and is severely mental impaired because of it, doctors tell us their is no treatment nor cure for it. After finding out about this disease, our doctors explain that not much is known about it because of its rarities found your web pages of which I copied and sent to all of her doctors. They were very interested in the information and reported to us that your information was more than they were able

to find out until now. Because of its rarity and because most doctors have not had any patients with it, not much effort was made to learn about it from the medical field. I just wanted to thank you for your work on this with your web page, and was wondering if any more information has been learned in Europe about this? Are they attempting to treat this any way? Its too late for help for my wife as she suffers from silent strokes and severe dementia, after having a brain biopsy to find out what she had. Any information you may have in addition to what you have on the web site would b e great. After reading your site and using this information to find out more on it, I printed this information and send it to my wife doctors. They were excited to get is as will help them with other cases that may come to them. Again thank you!