



# CADASIL Together We Have Hope Non Profit Organization www.cadasilfoundation.org Official Newsletter

Cerebral Autosomal Dominant Arteriopathy with Sub-cortical Infarcts & Leukoencephalopathy

## INFORMATION SHARING

Volume 8, Issue 1



March 2006

### NEW ONLINE FORUM FOR THE FOUNDATION

There is an online forum for everyone. Please free to post questions on this forum. Please go to [www.cadasilfoundation.org](http://www.cadasilfoundation.org), click on the red bar that says *forum* or go to <http://www.phpbbcity.com/forum/?mforum=cadasil>. It is intended to be a networking resource for sharing information relating to CADASIL, specifically to help others in a positive way.

It should not be used for advertising of services, personal gain, or marketing other commercial services. It is important to understand that this forum in no way constitutes medical advice. Your choice of treatment or whether you receive treatment is your personal decision. On the forum there are subjects like: frequently asked questions: how to find info on social security: recommendations, *L-Arginine*, ideas for raising awareness, and personal stories about the disease.

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<b>Special points of interest:</b>	
CADASIL Together We Have Hope was established on May 10, 2005, as a Non Profit Organization and a public charity.	

### FIRST TELEPHONE CONFERENCE FOR THE FOUNDATION

February 5<sup>th</sup>, 2006 was the first teleconference call, this will be a regular planned event. The discussion included symptoms of CADASIL, up to date research information, and a question and answer session. Please keep checking on the website for the next conference call date and time. The sessions will usually be held Sundays. The cost of the conference call is the domestic long distance rates charged by your long distance carriers for the length of the call. The conference call will last 30 minutes to an hour depending on the subject. The dial-in number is (605) 772-3001 and when asked, please use access code 836339#.

### INFORMATION FOR YOUR DOCTOR

We now have a separate page, go to website and click the button on Health Care Professionals. Please let your doctors know about this page as it contains the following: PowerPoint presentation on understanding CADASIL, MRI films and reports, blood test results, skin biopsy report and understanding vascular disease.

### WHY SEE A GENETIC COUNSELOR?

Knowing that you have a genetic syndrome (CADASIL) in the family means that you have many things to consider along with the medical implications and your reactions to a new situation. You may want to think about telling other family members, and letting them know how they could be tested to see if they also have the gene. If you have children, you will want to think about how having the syndrome could affect them, both emotionally and physically.

There are other important issues to consider, such as health insurance and genetic conditions. Some people will recommend you purchase life insurance for everyone in your family, even infants, before you have any of them interested for hereditary conditions. Many questions can come up. If you are looking for a genetic counselor you may go to the National Society of Genetic Counselors as they have a website that will help you locate one in your area: <http://www.nsgc.org/resourcelink.asp>

## **UNDERSTANDING CADASIL**

The following **Q**uestion and **A**nswer may be helpful in understanding the research activity occurring at Harvard and the University of Michigan.

Excerpts from website:

<http://ghr.nlm.nih.gov/search/term=12589106?area=pubmed-id> )

Q. What is CADASIL?

A. CADASIL is caused by a mutation in the Notch3 gene on chromosome 19.

Q. What genes are related to CADASIL?

A. The Notch3 gene makes a protein called the Notch3 receptor protein, which plays a role in the development, function and maintenance of vascular smooth muscle cells. Mutations in the Notch3 gene leads to an abnormality in the Notch3 receptor protein that then builds up in the vascular smooth muscle cells. Accumulation of the abnormal Notch3 receptor protein is thought to cause the degeneration of these muscle cells, leading to the loss of function of blood vessels in the brain and heart.

## **UPDATE ON RESEARCH AT HARVARD**

*E-mail from Joseph F. Arboleda-Velasquez, M.D. dated February 5, 2006.*

During the past few months we have made very important advances towards the development of animal models of CADASIL using both flies and mice. As you know, animal models of human disorders are an extremely important tool to better understand the pathological mechanisms of disease. Moreover, animal models are also essential to determine the safety and effectiveness of drugs. Specifically, we have generated mice carrying either a normal human Notch3 gene or two different Notch3 genes carrying specific CADASIL mutations. In addition, we are currently performing pathological and biochemical analyses of human brain samples from deceased CADASIL patients. The goal of these studies is to determine the composition of the granular osmiophilic deposits (GOM) that accumulate in the arteries of patients suffering CADASIL. As you know the characterization of brain deposits in other diseases such as Alzheimer's and Parkinson's has provide crucial insights

## **RESEARCH AT THE UNIVERSITY OF MICHIGAN**

Dr. Wang's lab is researching CADASIL as it is caused most commonly by a single point mutation in the Notch3 gene, which leads to alteration in the number of cysteines. (cysteines are amino acid that can be found in many proteins throughout the body) The lab is currently studying the mechanism of how mutant Notch3 leads to cellular injury. . He is actively pursuing the mechanisms of how the Notch3 gene is regulated, which may lead to ways to control Notch3 protein levels, which are typically elevated in CADASIL patients.

*Excerpt from an e-mails from Dr. Wang March 16, 2006, to the foundation* Thanks so much your invitation to describe more information on our research. We are doing well here; we received our first NIH grant to study the

Q. What mutations are effected with CADASIL?

A. More than 70 mutations that cause CADASIL have been reported. Almost all of these mutations change a single amino acid (a building block of proteins) in the Notch3 receptor protein.

Other known mutations have been reported: R142C, R133C, R170C, R90C, R182C, R169C. R141C, R110C (too many too list)

Please if you know your mutation contact us 1-877-519-HOPE or e-mail us at

[info@cadasilfoundation.org](mailto:info@cadasilfoundation.org) to share this information with researchers and the health care professionals.

Information contained in this newsletter is not intended as a substitution for professional medical care. Always check with your physician.



into the path physiological mechanisms of these disorders. In a similar way, the characterization of the GOMs may provide us with clues regarding the mechanism through which Notch3 mutations lead to CADASIL. I want to reassure you our team of scientist is doing everything in their hands to better understand how CADASIL works and how to treat this devastating disorder. I am very impressed to see the progress you have made in promoting awareness of CADASIL using the Foundation's website. I also appreciate your posted information about our laboratory (Dr. Artavanis-Tsakonas laboratory at Harvard University) in the research section of your website. <http://artavanis-tsakonas.med.harvard.edu/SATResearch.html>

molecular basis of CADASIL, and the team is working extremely hard (one of my post-doctors nearly lives in the lab). hope that these studies will eventually focus attention on potential. Excerpt from an e-mails from Dr. Wang March 16, 2006, to the foundation. Thanks so much your invitation to describe more information on our research. We are doing well here; we received our first NIH grant to study the molecular basis of CADASIL, and the team is working extremely hard (one of my post-doctors nearly lives in the lab).



### **CADASIL STUDY ENDING SOON**

The CADASIL Study is scheduled to end around May of this year; it is not too late to participate. The findings could be a break through to our families and the future of the next generation.

This study enrolls patients with a diagnosis of CADASIL that has been confirmed either by genetic testing or by a skin biopsy. To be eligible to participate, a patient should also:

Have some trouble with thinking or remembering [but this trouble need not be severe];

Have a spouse or other person who sees them regularly and can attend the evaluations with him/her [this allows the investigators to get another person's perspective on how the patient is doing.]

The study involves two screening visits, after which participants are randomly assigned to receive either a drug that improves cognition or a placebo for 18 weeks.

The participants return for regular follow-up visits at six, twelve and eighteen weeks. All visits involve interviews/questionnaires on symptoms and overall functioning, plus physical and neurological exams.

The above is a general description. Every patient with CADASIL is different, so interested patients or family members should contact the nearest center listed on the website to obtain more detailed and specific information or call 1-877-519-HOPE. Travel reimburse-

### **PUBLIC AWARENESS**

#### **INCREASE AWARENESS OF CADASIL WITH THE AMERICAN STROKE ASSOCIATION WEB SITE**

CADASIL Together We Have Hope is asking the American Stroke Association (a division of American Heart Association) to provide information about CADASIL on their website. To assist our Foundation please go to their website at <http://www.strokeassociation.org/presenter.jhtml?identifier=1069> and complete the information on the requested form:

On the question 'I would like to learn more about' enter CADASIL in the 'other' field.

The next question 'I heard about the American Stroke Association from' enter CADASIL Together We Have Hope Non-Profit Organization in the 'other' field

If you would like to write to the American Heart Association their address is American Stroke Association, National Center, 7272 Greenville Avenue Dallas TX 75231 Please remember to include our foundation in your letter.

This will help facilitate the process.

#### **NORD CONFERENCE REPORT SEPTEMBER 2005**

In September 2005 one of our board members represented the foundation at the National Organization of Rare Disease meeting in Rockville, Maryland. The conference addressed the growing public need for improvement in availability, and quality of genetic and other diagnostic laboratory testing for rare diseases and conditions. This was the second conference. Breakout sessions were focused on addressing these issues and the consensus was a need for improved testing. It was interesting to hear other genetic disorders being presented.

During the conference Sharon Terry, Director of the Genetic Alliance introduced us to a lively cheerful Congresswomen Judy Biggert from the 13<sup>th</sup> District of Illinois. Ms. Biggert talked about her role in different aspects of congress then discussed a genetic nondiscrimination bill. She addressed the problem we have with the genetic nondiscrimination act today; as genetic information becomes increasingly integral to the standard practice of medicine, the need for legislation that protects against genetic discrimination becomes absolutely essential. Currently, the US House of Representatives is considering a bill that would provide that protection. The Genetic Information Nondiscrimination Act of 2005 (S.306, H.R.1227), which received unanimous approval in the Senate earlier this year, will ensure that both issuers of health insurance policies and employers provide health insurance coverage without evaluation of genetic information. As well, employers make cannot make employment decisions

based on an individual's genetic information. Today, opposition to this critical legislation comes, primarily, from employment trade organizations that claim that genetic discrimination is not happening. Unfortunately, these organizations are wrong; genetic discrimination is a very real phenomenon. Considerations in testing family members. Why we should care, examples of who is impacted and how The bill introduced to Congress Activity in Congress to date on the bill

1) **What you can do:** Considerations in testing family members: *CADASIL Together We Have Hope* receives e-mails from families wanting to have their family members tested but due to genetic discrimination are scared about the future; our own family is affected with this discrimination.

The voices of a few should not dominate this discussion. Cases of discrimination, and the very real concerns associated with the possibility of discrimination, are already affecting the quality of our health care and clinical trials.

2) **Why we should care:** examples of who is impacted and how: The current patchwork of state laws leaves individuals vulnerable - There are a few states with strict protections against genetic discrimination, but most states have little to no protection. This leaves individuals with little knowledge about how much their genetic information is protected from state to state. In addition, companies who deal in any kind of health information are left with no national framework to guide how they handle genetic test results and genetic privacy information.

Genetic information is not properly covered under HIPAA privacy guidelines. HIPAA or Health Insurance Portability and Accountability Act was created in 1996 to help secure families with health insurance. In a day and age when we change jobs as much as our clothes, HIPAA has come along to help people carry health insurance throughout their job transitions. Thus the "Portability" in Health Insurance Portability and Accountability Act (HIPAA). Current HIPAA guidelines do not prohibit insurers from requiring genetic testing or from denying coverage based on genetic information. In fact, GINA (Genetic Information Nondiscrimination Act) is filling holes in HIPAA, making the whole legislation more consistent with regard to data protection. Genetic information is becoming increasingly more ingrained in medicine and as such will serve to complicate privacy law if steps are not taken now to close gaps in policy.

Genetic discrimination cases are already appearing and are sure to exponentially increase with so many new genetic tests coming out.

#### **For Consumers -**

Your genetic information could be used to deny, limit, or cancel your health insurance. You could face genetic discrimination in the work place.

**For Health Professionals** - Patients are refusing beneficial genetic tests due to fear of misuse of genetic information.

**For Researchers** - Genetic discrimination fears are slowing clinical trials and decreasing the number of effective treatments being developed.

#### **For Employers-**

the lack of a framework for genetic privacy increases the risk of unregulated legal action.

3) **The bill introduced to Congress:** Genetic Information Nondiscrimination Act of 2005 - Amends the Employee Retirement Income Security Act of 1974 (ERISA) and the Public Health Service Act to expand the prohibition against discrimination by group health plans and health insurance issuers in the group and individual markets on the basis of genetic information or services to prohibit: (1) enrollment and premium discrimination based on information about a request for or receipt of genetic services; and (2) requiring genetic testing. Sets forth penalties for violations. Amends title XVIII (Medicare) of the Social Security Act to prohibit issuers of Medicare supplemental policies from discriminating on the basis of genetic information. Extend medical privacy and confidentiality rules to the disclosure of genetic information. Makes it an unlawful employment practice for an employer, employment agency, labor organization, or training program to discriminate against an individual or deprive such individual of employment opportunities because of genetic information. Prohibits the collection and disclosure of genetic information, with certain exceptions. Establishes a Genetic Nondiscrimination Study Commission to review the developing science of genetics and advise Congress on the advisability of providing for a disparate impact cause of action under this Act .

4) **Activity in Congress to date on the bill:** 109th Congress Activity

March 10, 2005: H.R. 1227 introduced to the House and referred to the Energy and Commerce, the Education

and Workforce, and the Ways and Means Committees. (Over 120 cosponsors)

Feb 17, 2005: S. 306 passed in the Senate, 98 – 0.

Feb 16, 2005: President Bush issued Statement of Administration Policy , supporting S.306.

Feb 7, 2005: S.306, Genetic Information Nondiscrimination Act, introduced

History of the Act in the 108th Congress

October 1, 2003 – The full U.S. Senate voted unanimously (95-0) in favor of the bill, and President Bush issued a Statement of Administration Policy in support of the bill.

May 2003 – Senator Olympia Snowe (R-ME) introduced bill S.1053 "Genetic Information Nondiscrimination Act of 2003"

The bill did not move in the House throughout the 108<sup>th</sup> Congress.

Genetic Information Nondiscrimination Act [H.R. 1227, S. 306]

**5) What you can do:** Visit your representative today or send a letter to your representative to let them know you expect them to act on H.R.1227. Reference: Coalition of Genetic Fairness web site.

## **ANNOUNCEMENTS HOW YOU CAN HELP AND HELP OTHERS**

### ***First Anniversary!***

*CADASIL Together We Have Hope celebrates its first anniversary May 2006.*

We will be having our first annual fundraiser to celebrate our anniversary. Please check for information on the website, in the mail, as well included in the next newsletter .

### ***Recognized by the Internal Revenue Service as a Public Charity***

We are pleased to announce that *CADASIL Together We Have Hope* is now a 501 (c) (3) public charity recognized by the Internal Revenue Service. What this means is that all donations are tax-deductible. We are also qualified to receive tax-deductible bequests, devices, transfers or gifts. *Together We Have Hope* is making a difference in the lives of people touched by CADASIL. Contributions will be used 100% towards making our mission and our vision become a reality. We are beginning to write grants for a CADASIL Conference to be held here in the U.S.A. in the spring/summer of 2007.

### ***The foundation currently is:***

- Mailing information packets weekly to those recently diagnosed and sending information packets to their doctors to educate the doctor on CADASIL.
- Organizing and planning to hold the first symposium on CADASIL in the U.S.A
- Identifying physicians and neurologist to volunteer to be patrons of the Foundation
- Establishing a scientific board to educate doctors, radiologists, neurologist, and directors of strokes, and others who may be presented with a person with symptoms of CADASIL, but fail to recognize them as such.
- Disseminating and sharing the latest information through the internet website (e.g., information, studies, testing sites, news articles, newsletters, research, doctor pages, and confirmed cases)
- Providing awareness on the disease through conference attendance and sharing of information by reporting the outcome of the attended conferences. Working closely with a production specialist to inform physicians about CADASIL
- Members of the American Brain Coalition, which is comprised of some of the United States' leading professional neurological and psychiatric associations and patient organizations. Through involvement we seek to understanding the functions of the brain, and to reduce the burden of brain disorders through public advocacy)
- Members of The Coalition for Genetic Fairness, which was founded in 2000 to address the growing concern, surrounding the misuse of genetic information in insurance and employment decisions. The Coalition's objective is to urge Congress to pass comprehensive federal legislation outlawing genetic discrimination and to educate the public about these protections
- Recognized all over the world by leading experts on CADASIL

**Make sure you have signed our guest book or call us at 1-877-519-HOPE**

Please provide us your information so we can mail you a welcome packet. Once you have registered with us, your information is transferred to a database, which tracks the number of confirmed cases all over the world. You will be kept informed with up-to-date information about CADASIL by receiving e-mail alerts and newsletters in the mail. The foundation does not require membership to join. All personnel information provided to us is **kept strictly confidential** <http://home.earthlink.net/~cadasil101/>

**Creating an Emergency Care Plan**

Developing an emergency plan is very important, be prepared and plan today. Go to the website and click on emergency plans and print out the business size emergency card for your wallets and family members.

**Charity Shopping Network**

Do you shop online? Donate by shopping online, with charity shopping networks! Shop online and the vendor's donation a portion of your purchases to the CADASIL Together We Have Hope (Foundation). If you already shop online with retailers such as Best Buy, Expedia, Home Depot, Old Navy, E-Bay, Dell Computers, Sears, Target, and many, many more, then why not have a portion of your purchases help us.

Your loyalty helps raise money for CADASIL, without costing you anything extra... not even a penny. There are hundreds of companies across a broad range of categories, which want to support you if you support them. By shopping at participating companies, your purchases can have a positive impact on CADASIL and helping to raise funds, which will be, used 100% towards our mission. Website located @



[www.igive.com](http://www.igive.com) or <http://www.tricor>

**PERSONAL STORIES****Family Tree Loses Genetic Defect (CADASIL)**

Before you read this article, I would like to define what is pre-implantation genetic diagnosis is - This is a procedure that eliminates genetically defective human embryos before they have a chance to develop. Prospective parents who are concerned about passing an incurable genetically based disease or disorder to their child usually request this. Typically one or both partners have been genetically screened previously, and found to be a carrier.

**This following is excerpts from the Charlotte Observer, June 20<sup>th</sup>, 2005. Due to the Brinson's privacy I have not included their first names.** *From an article written by: Karen Garloch*

A 40-year-old Charlottean learned three years ago that he had CADASIL. Except for an especially bad episode three years ago, Brinson has controlled his condition by treating the symptoms. Looking back, he realizes he inherited the condition from his father, who died in his 60s, and that his father inherited it from his own mother, who died in her 80s. Anyone with the condition has a 50-50 chance of passing it to a child. And, Brinson and wife have been told that as it passes through each generation, symptoms can start earlier.

He had been married nine years, and they had talked about having children. Once they got the diagnosis, they concluded they would have to adopt. But last summer, Mrs. Brinson tried one last time to find a way to have a biological child.

She knew there were tests for abnormalities after conception, such as chronic villus sampling and amniocentesis. By searching the Internet, She learned about pre-implantation genetic diagnosis, or PGD, a test that can detect a single-gene defect in an embryo before it is implanted into the uterus during in vitro fertilization (IVF). She called a reproductive endocrinologist in Charlotte, Dr. Gordon Kuttner, who agreed to help them. He contacted Dr. Mark Hughes, director of Genesis Genetics Institute. Hughes had developed PGD more than a decade ago, and Kuttner knew it could detect genetic abnormalities, such as cystic fibrosis and sickle cell anemia. It had never been used to test embryos for CADASIL, Genesis officials told Kuttner. But they were able to create a specific marker to identify CADASIL.

To get the embryos, the Brinson's started a cycle of IVF, a technique usually reserved for couples that cannot conceive naturally. Nine embryos lived to be three days old, and the lab tech took single cells from each and sent

them to Genesis. For the next 19 hours, a team of Genesis scientists tested the embryos and found four that did not have the defective gene. They relayed that information to Kuttner. Two of the four healthy embryos were transferred into Mrs. Brinson's uterus on Day 5. The other two were frozen for possible future use. The unhealthy ones were discarded. That was Nov. 20. Mr. Brinson, 40, and his wife, 35, are expecting their baby the first week of August.

Although they know some people would question the ethics of discarding embryos, the Brinsons are glad they found a way to rid their family tree of a serious genetic problem without having to face the decision to abort a fetus later in development. The Brinsons believe, as does Kuttner, that they practiced "preventive medicine". "We found out we could effectively knock this out of the family tree, so we've done that," Mr. Brinson said. "There's a tremendous relief in knowing that."

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Follow up after the story -

One of our directors, Billie Duncan-Smith had the pleasure to contact Dr. Mark Hughes and this is what his reply was. "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. The cost for IVF varies GREATLY across the nation. The actual Pre-implantation genetic testing could be paid from different types of grants and foundations. And, keep up the great work! Families need organizations and information websites like this! Congratulations." Mark R. Hughes, MD, PhD, Director, Genesis Genetics Institute, Genomics Center Samartan

### ***Coping with a CADASIL Life***

***By Sarah Moris***

As we all know, coping with life's circumstances while dealing with the circumstances and symptoms that CADASIL brings us everyday can be a daunting and overwhelming challenge at times. We know this because we can assume that if you're reading this letter you may have CADASIL, or know of someone who does. Or perhaps you know of someone who has the genetic mutation for a disorder, but remains symptom-free, as is my case. I am almost thirty years old and was made aware of CADASIL in our family just last year. I knew right away that I wanted to be tested for the mutation, but yet took some time to be sure that it was the right decision. And while I did question whether or not it was the right thing to do at times, I never did change my mind. I wanted to know. While the test results were positive and it wasn't the news that I wanted to hear, I immediately felt relief in knowing. However, I do know that predictive genetic testing is not for everyone. We all have different coping mechanisms in dealing with life's circumstances.

But whether you know you have CADASIL or not, or whether you know you might have CADASIL or might not, the bottom-line is that we all have to cope with this. We all have lives to live. Our friends and families deserve to have the best of us. Before getting tested, I made the conscious decision that these test results were not going to control my life. If that was going to be the case, then I should've never have been tested.

I believe that I am coping with CADASIL in my life today pretty well. I know I am dealing with it much better than I was last year, and I hope that I'll be doing better next year than I am now. But there were certain steps I took to get myself to where I am today. First of all was seeking the medical help that I so desperately needed after a positive diagnosis. The fear and anxiety were overwhelming, and finding a good Psychiatrist (a doctor who prescribes medications) and a Psychologist (a Therapist) was essential to getting me well again. It was the combination of medications and therapy that allowed me to start to feel like myself again. The amazing part was that I never realized how bad I felt until I started to feel good again. And I hid it well. I know that even my closest friends and family had no idea how hard it was for me to get out of bed some mornings. Or I could tell even my own small children were happy to get their playful mother back again, instead of this tired, irritable, anxiety-ridden woman pretending to be their same mother.

The next pivotal step for myself was "finding my faith." I had so many questions, but I never asked "Why me?" or "Why my family?" I do believe that I was given CADASIL in my life for a very specific reason. And even though I don't understand the reasoning, or what I'm supposed to do about it, I know that I will understand someday. I also believe that the timing in which I discovered CADASIL in my life was meant to be. My father was tested seven years earlier, before there was a DNA testing method available. He had a brain biopsy performed, and the results were falsely negative. It was not until last year that his primary care doctor heard of a DNA test available and thought we should test again, even though there was any family history of early-onset

stroke or dementia. For myself, I personally know that I would've never have had children had I known about CADASIL ahead of time. It amazes me that I have these beautiful children in my life, and it baffles me to think of my life without them. Therefore, I know these children were meant to be. Even though they're already amazing little people, I know they're going to grow up to be something great someday. A friend of mine was giving an analogy of life and happiness to that of flying an airplane. Every airplane needs two things to fly: "thrust" and little "drag" or "resistance". The tiniest little screw that can create drag against an aircraft can significantly reduce its efficiency. We all deserve happiness (i.e.-"thrust") in life. What is it in your life that is your thrust? Whatever that is: embrace it! What is it in your life that is your drag? Whatever it is: diminish it! You can't soar in life until you diminish your drag. At that moment my friend was telling me this, I realized my fear was my drag. I couldn't soar or obtain happiness until I diminished my fear. My fear of what 'might be some day' was robbing my family and me of my daily dose of happiness. This friend gave me two assignments to absolve my fear. First was to think of whatever it was that I feared, in other words-the worst possible outcome that I could think of, and tell yourself that if that happens to me: IT'S OKAY. Whatever it is, everything will be okay because it was all meant to happen. There will be a lesson as a result of it. The second assignment was to stop with the negative thinking. For every negative thought I worried about disability, dementia, or any of the symptoms of CADASIL could bring, I had to think a positive thought. For me, any thought I had of my father's disabilities and wondering if I would suffer from the same limitations, I pictured myself as a perfectly healthy 90-year-old grandmother enjoying her grandchildren. Before my friend had given me this assignment, I was ashamed to admit that I hadn't even done this once since I got my test results! I had taken the information from the geneticists and Internet and thought this is the way it will be for me. But I realize now, that nobody can predict the future. And I also know now that even if that is my fate, it was meant to be.

In a strange way, CADASIL has enhanced my life. My priorities are crystal clear to me now. But I won't pretend for one minute that I still have my good days and bad days. But it is because I have surrounded myself with people who are supportive, positive, optimistic and encouraging to rely on when I need it most that I can get thru those bad days just fine. I don't want people's pity. Do I want compassion and understanding, absolutely! But pity won't get me anywhere in life. Negativity, fear and worry won't get me anywhere, either. I'm at a good place with my life now. I feel that I've reached the "land of acceptance", but it's been a journey. And there is no doubt in my mind that the journey will continue. Life is what you make of it, and I look forward to the journey that I have in my future.

### ***A CADASIL Diagnosis – What and How it Happened*** ***Anonymous***

"The trial lawyer who could not talk", that was how the doctors referred to my husband's case. My husband was recently diagnosed with CADASIL. Fortunately we live in Chicago and have access to excellent hospitals that knew about CADASIL. We sought opinions from three different doctors from three hospitals. Doctors at two of the hospitals began to collaborate on his case. It took a total of 5 weeks to diagnose my husband.

His symptoms presented through aphasia; knowing what he wanted to say, though the inability to find the words. He experienced two episodes one evening, a few hours apart lasting only a few minutes. We were fortunate that when we made the phone call to our general practitioner he encouraged us to go the emergency room immediately. It was there that a CAT scan was done and inflammation was seen in the brain. At about midnight doctors were brought in to do an MRI and demyelination of white matter was identified throughout much of the brain. It was thought the most likely cause was MS and treatment of high levels of steroids was done; very important to note, steroids should be avoided with CADASIL. The doctors considered a number of diseases in the diagnosis, and my husband was tested for each: Multiple Sclerosis, ADEM, Infections, various autoimmune, Vasculitis, Mitochondrial, Leukoencephalopathy, and CADASIL. CADASIL at the time was considered unlikely because both of my husband's parents were in their 60s with no symptoms. It was the Athena genetics lab test that provided positive diagnosis for CADASIL. After diagnosis, both of my husband's parents were tested. His mother was diagnosed with CADASIL at age 61 with no symptoms. My husband's grandfather (his mother's father) was diagnosed and died from NPH at age 60 and his great grandfather had a low pressure stroke; which now we know was misdiagnosed many years ago. Our concern now is our 6-month-old son. Each day we hope and pray he does not have CADASIL.

Thank you for reading this article.

**Please send us your stories. (Remember to keep them brief)**

### Newsletter Word Search Challenge

Find all of the words which are located in this newsletter . Words are either forwards, backwards or diagonal.  
Have fun!

N	C	L	P	N	A	U	T	N	A	A	U	E	N	S	G
O	G	Y	T	S	S	C	O	A	W	R	G	E	N	E	S
I	E	N	S	R	N	O	I	T	A	D	N	U	O	F	I
T	N	O	I	T	A	T	U	M	R	S	I	N	N	S	O
A	E	G	G	P	E	T	O	G	E	T	H	E	R	G	W
N	T	V	O	U	P	I	G	G	N	N	C	C	D	H	P
I	I	A	L	O	E	O	N	S	E	A	R	I	I	D	P
M	C	S	O	R	A	S	H	E	S	R	A	T	D	S	U
I	I	C	R	G	S	O	T	S	S	G	E	S	Y	O	R
R	O	U	U	R	L	I	O	B	N	B	S	Y	T	S	N
C	S	L	E	N	A	C	A	O	O	R	E	M	G	I	S
S	L	A	N	O	I	S	S	E	F	O	R	P	E	T	E
I	I	R	T	A	H	I	P	P	A	T	K	T	R	N	O
D	P	N	T	R	S	T	U	D	Y	M	O	O	N	A	P
N	O	I	S	S	A	P	M	O	C	R	K	M	G	O	P
O	O	T	I	I	O	S	O	O	P	E	O	S	M	N	A
N	N	O	S	S	R	D	P	O	O	G	H	C	W	R	O

<input type="checkbox"/>	association
<input checked="" type="checkbox"/>	cysteines
<input type="checkbox"/>	genes
<input type="checkbox"/>	guestbook
<input type="checkbox"/>	neurologist
<input type="checkbox"/>	protein
<input type="checkbox"/>	stroke
<input type="checkbox"/>	together

<input type="checkbox"/>	awareness
<input type="checkbox"/>	diagnosis
<input type="checkbox"/>	genetic
<input type="checkbox"/>	HIPPA
<input type="checkbox"/>	nondiscrimination
<input type="checkbox"/>	researching
<input type="checkbox"/>	study
<input type="checkbox"/>	vascular

<input type="checkbox"/>	compassion
<input type="checkbox"/>	foundation
<input type="checkbox"/>	grants
<input type="checkbox"/>	mutation
<input type="checkbox"/>	professionals
<input type="checkbox"/>	shopping
<input type="checkbox"/>	symptoms
<input type="checkbox"/>	white

If you need the answer key, e-mail us @ [info@cadasilfoundation.org](mailto:info@cadasilfoundation.org).



CEREBRAL AUTOSOMAL  
DOMINANT ARTERIOPATHY WITH  
SUB-CORTICAL INFARCTS &  
LEUKOENCEPHALOPHY



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HAVE HOPE

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WE'RE ON THE WEB!  
[www.cadasilfoundation.org](http://www.cadasilfoundation.org)

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*IN LOVING MEMORY OF*

*Charlie Charron of Sunbury, Ontario, CANADA - Our thoughts and prayers are with his wife, Anne and their family.*



*J. Richard "Rudy" Ruedebusch of Plover, Wisconsin, USA - Our thoughts and prayers are with his wife, daughter Sara Moris and family.*

*Please contact us if you would like to add a memorial to our next newsletter.*

Thank you for those who contributed to this newsletter

*Billie Duncan-Smith, Director*  
CADASIL Together We Have Hope

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