IRS 501(c)(3) Approval Granted!

On September 24, 2012, CADASIL Association, Inc. was approved as a non-profit public charity, retroactive to the date of the association’s incorporation of April 24, 2012. All charitable donations since April 24th are tax-exempt and may be claimed as tax-deductible contributions.

Please join us by becoming a member of CADASIL Association! Membership is free and open to anyone touched by or interested in the rare genetic disease CADASIL. We would also love to put your talents to work. Let us know how you can help CADASIL Association meet the goals in our mission statement. Go to: www.cadasilassociation.org/Join_Us_2.html

Want to acknowledge someone in the CADASIL community with a contribution in their honor? Tax-exempt donations make wonderful gifts for holidays, birthdays, and other special occasions. You can help our mission of Communication, Advocacy, Research, and Education by donating to CADASIL Association at www.cadasilassociation.org/Donate.html

Want to tell your story? Have a question for the doctor? Planning an event? Upcoming newsletters in 2013 will feature CADASIL patients and their loved ones sharing personal stories of living with CADASIL or caring for someone with this rare genetic disease. We welcome your submissions of stories, questions, news, and events. Please email them to: embracenewsletter@gmail.com
I never had headaches or migraines growing up, so when I started getting them when I turned 40 years old, I thought that was strange. I started seeing a neurologist and had an MRI. The neurologist told me I might have Multiple Sclerosis but he wasn’t sure. He told me it was my choice on whether to go on MS medications or not. I chose not to. He began treating me for migraines.

One day at my job, while just standing and talking to a co-worker, it felt like a sledgehammer hit me in the back of the head. I was escorted to my desk to sit down. I could only stare. I could not talk or move. This felt like it went on forever. I finally came back to consciousness while a paramedic was talking to me in the ambulance. At the hospital they really did nothing for me, and I was released a couple of hours later. At home, I continued to have frequent “staring episodes” followed by migraines. I decided to have someone take me back to the emergency room. My episodes seemed to baffle the doctors. They sent me for a MRI, and after that they admitted me for more testing.

My father had been diagnosed with dementia when he was in his late forties/early fifties. He had a neurologist at this same hospital, and when he passed away at the age of 58 we allowed that hospital to do an autopsy. (Hence the reason I chose this particular hospital.) A neurologist came to my room after looking at both my MRI and my father’s autopsy report, and sat down to tell me he thought I have something called CADASIL. He told me about it and asked for approval to do the genetic test. I was sitting there with my mom and my best friend. As the doctor was explaining all this to us, I looked over at my mom and she had tears in her eyes. As for me, I couldn't believe it, and I don’t even remember much of what he said or if I was really listening to him.

A month later, my husband and my mom and I went to hear the results of the genetic test. As I sat there waiting, I felt really nauseas and anxious. When the doctor came close to me and said, “The blood test came back positive, and the skin biopsy also came back positive,” I think I was in shock. Again, my mom had tears in her eyes. I can say that in some weird way I blamed my dad for this, which is really stupid of me for he didn’t know what was wrong with him. None of us knew. I got very depressed and felt a wave of doom come over me. I thought back how bad my dad’s condition got and how quickly it all happened to him. I kept thinking I’m going to die in 5-10 years. I thought how my mom had to deal with my dad at his worst, and in some ways I didn't want my husband to have to deal with me at my worst.

Now, over two years after my diagnosis, I still get bouts of major depression, but I'm finding that I have to live my life to the fullest no matter what. I have my good days and my bad days, but my CADASIL is not going away, so I have to live with it and be as hopeful that I can that there will be research done and a cure found.

I meet more and more people online at Facebook who have CADASIL, or who know someone who has it, or who have had a loved one pass away from it. There has to be even more people out there who have CADASIL and just don’t know it yet because their doctors haven’t heard of it.

My goal is to get the word out in any way possible. **KNOWLEDGE IS POWER!!**
My first experiences with persistent headaches were in high school. I have memories of occasional headaches during childhood and early adolescence, but those that started when I was around sixteen could be brutal. I had been having them for weeks and missing a lot of school when my dad took me to his chiropractor. X-rays showed I had various vertebrae out of whack. The treatments helped, but I sometimes felt more nauseous and dizzy after a spinal adjustment than before. I didn’t tell my parents this because they were relieved to think I was getting help.

In my twenties and thirties I rode the joyous ups and complex downs of being married and raising a young son, while working many hours in a hospital office and taking college classes toward becoming a teacher. I clearly recall battles with depression, anxiety, mood swings, and fatigue, but I assumed those went along with my hectic life. I hid these symptoms behind a smiling face and a good sense of humor. I attempted to keep my headaches at bay with large doses of non-prescription pain relievers. I’d do anything to numb the insidious pain.

A divorce in my late thirties took me as low as I believed I’d ever be. The failure of my marriage hit hard, as I had always been driven to succeed in everything I held important. My son, by then a young teen, was hurting and extremely angry. I felt like a rotten mother and a fragmented shell of the person I once knew.

Meeting my second husband brought newfound happiness. I still had headaches, but my depression lifted for a while as I settled into a good marriage. My relationship with my husband remained strong, yet the depression and other issues returned. I did not understand why I could feel so awful at times. My counselor referred me to a psychologist, and I began more intensive talk therapy and opted to try anti-depressants. I could feel some relief, but I knew I still wasn’t well. I feared I might be bipolar, although my psychologist assured me I was not.

Through the summer of 2008 I went on and off several medications during my vacation from school to try and balance my emotional swings. Surely the drugs caused the now frequent sick headaches, I thought. As autumn approached, I felt so crummy that I seriously considered ending my life. I became increasingly ill, barely managing to rally for my job only to collapse at home later. Daily I had tight pressure across my forehead into the top of my skull. Sometimes if felt as though an invisible hand was twisting my brain, and I would get waves of nausea and dizziness. I kept the truth of how terrible I felt from my husband, my other family members, my friends, and my colleagues. I feared I was going insane, and that if anyone knew how sick I was, I would be institutionalized.

Near the end of October 2008, I blacked out while attempting to get ready for work. I called my husband when I regained consciousness, and he rushed home to take me to the hospital. (Please also see my story regarding my ER visit and my neurologist on p. 5.)

Being diagnosed with CADASIL nearly a year later, a month before my 50th birthday, was devastating. How could I have a rare genetic disease few people know exists? At the same time, my diagnosis explained many of my symptoms and answered a lot of questions. In a way, knowing I have CADASIL has helped me cope with my life. Certainly, it has made me appreciate my good days more fully, and it has allowed me to give myself permission to take it easy when I am feeling sick.

My diagnosis renewed my faith in God. I work at maintaining a positive attitude and keeping myself as healthy as I can even while I realize this disease is progressing in me. I have hope for a cure for CADASIL – if not in my lifetime, then in the lifetime of the next generation.
ASK DR. JOE ARBOLEDA

Special to this inaugural issue of
EMBRACE:
Getting to Know Dr. Joe Arboleda

Dr. Joseph F. Arboleda-Velasquez was introduced to the field of study of SVD (small vessel diseases) as a medical student in Colombia, South America. He worked under the supervision of internationally recognized neurologist Dr. Francisco Lopera in the clinical characterization of two large Colombian families with CADASIL. Joe (as he prefers to be called) has had his clinical work reported in several research publications.

Joe came to the USA in the year 2000 with the goal of becoming a scientist. He has worked in laboratories and programs at Brigham and Women's Hospital and at Harvard Medical School. He joined the Schepens Eye Research Institute in 2010 as a post-doctoral fellow to study the role of cell-cell interactions in diabetic retinopathy. Joe was appointed as Faculty Investigator at Schepens in 2012 to begin his independent scientist career. His current research includes cell signaling and the pathobiology of small vessel diseases (CADASIL). Please view his professional website at: http://www.schepens.harvard.edu/arboleda

Joe is married and has a young son named Manuel Jose. He likes reading, writing, and practicing yoga.

Karla Smith recently interviewed Joe:

How did you first learn about CADASIL? While I was in medical school my research mentor introduced me to a family that he thought had CADASIL because they suffered from stroke and vascular dementia. I decided to work with this family as part of my internship to help them uncover the cause of the disease.

Why are you interested in CADASIL? I feel very committed to help the families with CADASIL because I know firsthand about their suffering and see how committed they are to help each other.

What kind of research have you done for CADASIL? I have worked on many aspects of CADASIL over the years. I started doing genetic work to identify mutations in Notch 3 associated with CADASIL. Later, I did some molecular work to study how CADASIL mutations affect the Notch 3 receptor. More recently, I developed animal models of CADASIL that can help us to deepen our understanding of this condition and test therapies.

What kind of research are you doing now for CADASIL? Now my work is focused on testing drugs that could help in treating CADASIL. Specifically, I am testing the effect of drugs that modulate Notch 3 activity on mice that suffer from CADASIL-associated changes. This work is now being conducted on laboratory animals to test for safety as well as efficacy.

Do you have any family members or friends who have CADASIL? My father was recently diagnosed with dementia. In many CADASIL patients, the multiple stroke events lead to dementia. My father's condition is stable now, but having my own father suffer from dementia helped me to get very close and identify with the CADASIL community.

Have you written any articles on CADASIL? I have published several articles on CADASIL and many others on the roles and functions of Notch receptors, one of which is affected by the CADASIL mutations. Writing is also my passion and thus I look forward to writing much more about CADASIL and help many patients this way.

If you have questions for Dr. Joe Arboleda, please send them to embracenewsletter@gmail.com
OUR DOCTORS

Karla J. Smith’s Neurologist: Dr. Yasushi Kisanuki, M.D., Assistant Professor, Department of Neurology, Division of Neurogenetics at Wexner Medical Center at The Ohio State University (Phone: 614-293-4969 / Fax: 614-293-4688)

When asked how he learned about CADASIL, Dr. Yasushi Kisanuki replied, “We learn CADASIL during medical school and neurology residency. I personally diagnosed patients with CADASIL first time when I was in training of neurogenetics (this is after my neurology residency training). CADASIL is frequently overlooked/misdiagnosed as ‘MS’. Due to the requirement of specific diagnostic tests (such as skin biopsy and/or genetic test), physicians can miss this diagnosis if they do not "suspect" it as differential diagnosis. Due to the risk of genetic inheritance, diagnosis of CADASIL can further influence familial dynamics, and genetic counseling is necessary before confirmation of diagnosis.”

From Karla: I was fortunate that Dr. Kisanuki happened to be working at Wexner Medical Center when I went to the emergency room there. He is the doctor from my story on page 2 who had an idea of what might be wrong with me. When he sat down to speak with me while my mom and my best friend were in my hospital room, Dr. Kisanuki mentioned the one word that would change my life forever: CADASIL. He explained what CADASIL is (we had never heard of it before) and what it does. My thanks go to Dr. Yasushi Kisanuki and his previous history of having CADASIL patients for being able to diagnose me as quickly as he did. He is a very kind and caring neurologist.

Janet Mills’ Neurologist: Dr. Angelo M. Santiago, M.D., Owner of Central Wyoming Neurology in Casper, Wyoming (Phone: 307-234-9037 / Fax: 307-234-9042)

From Janet: I met Dr. Santiago for the first time a few days after I blacked out in 2008 and my husband took me to the emergency room. From the images on my MRI at the hospital that day, the ER doctor and radiologist said it looked like I had MS. My regular doctor said “Not so fast” and referred me to Dr. Santiago. From the beginning of our now four-year doctor/patient relationship, Dr. Santiago has had knowledge of CADASIL. He said it was a possibility after viewing my MRI images at our first appointment. However, since CADASIL is genetic and neither my family nor I had ever heard of it, I didn’t follow up on his recommendation to see a specialist and colleague in Denver. Instead, I endured almost a year of other tests to definitively rule out MS and a number of other conditions.

When my husband and I did see the specialist, he agreed with Dr. Santiago that I could have CADASIL and ordered the test. When we returned to Denver for the results, I knew the moment the doctor walked into the exam room that I had this strange, little-known disease by how he didn’t tell me right away that I did not. When he said it out loud, the moment was surreal. (Please also read My Story on p. 3 of this newsletter for more about this experience.)

When I next saw Dr. Santiago, he expressed sympathy at my diagnosis. In a compassionate manner, he delivered the news that the disease would progress in my brain, and said he held hope for a slow progression of symptoms in my case. I credit Dr. Angelo Santiago for his knowledge of CADASIL and for his caring attitude toward me every time I see him.
2012 CADASIL Forum Conference

Five members of the Board of Trustees for CADASIL Association attended the CADASIL Forum Conference in Salt Lake City, Utah on September 12-14, 2012. While our Board meets via conference call at minimum once a month, this was our first opportunity to get together in person since the association was formed in the spring.

We learned a lot from the medical professionals at the conference organized and sponsored by the Russell family. Many thanks to them for a successful conference where many CADASIL patients and families could meet up and gain more knowledge about this genetic condition.

Visit the CADASIL Forum website at: http://www.cadasilforum.org

Speaking of conferences, be sure to check out our Upcoming Events on p. 7!
MORE RECENT NEWS

CADASIL Awareness Day

Members of CADASIL Association and the global CADASIL community participated in the third CADASIL Awareness Day on Friday, November 16th, 2012. Methods of participation included:

- sending information and/or a press release about CADASIL often being misdiagnosed as MS (Multiple Sclerosis) to newspapers, online bulletin boards, and Facebook pages
- emailing news organizations and TV programs with the request to feature a story about CADASIL
- posting CADASIL Awareness signs at local libraries, grocery stores and on other community notices boards
- requesting friends and family to name CADASIL Association as their non-profit organization of choice at Good Search/Good Shop/Good Dining at: www.goodsearch.com/nonprofit/cadasil-association.aspx
  (You can still sign up as a supporter at the above link!)
- telling one (or more) person about CADASIL
- spreading the word about CADASIL in various other creative ways

Ribbons (such as these to the left made by Trustee Freddie Schilling) are often worn or displayed by members of the CADASIL community to spread awareness about this rare disease.

Many people in the global CADASIL community use this image to bring awareness to this disease and to explain the words behind the acronym. Member Karl Stumpf has given permission for this image he created to be shared.

The article to the right was published in Trustee Janet Mills’ local newspaper, the Casper Star-Tribune, from November 17th to approximately November 24th.
UPCOMING EVENTS

Rare Disease Day 2013

Rare Disease Day, acknowledged by many countries around the world, is coming up again on February 28, 2013. Please check the website shown in the image above for ideas on how to honor this day in your community, state, or nation. Last year Trustees Anne McGuinness and Barbara Hunt attended meetings and workshops in Washington, D.C. in conjunction with Rare Disease Day. They hosted a table with CADASIL information at the NIH (National Institute of Health) Event Hall.

Trustee Janet Mills submitted the video “Faces of CADASIL” to the RDD website for 2012. It continues to be shown on their Videos page at: http://video.rarediseaseday.org/video-gallery, can be viewed at CADASIL Association’s home page, and is on YouTube at: http://www.youtube.com/watch?v=BmHUu7Wz5lc

Janet gives permission for anyone to share the video in its entirety

2013 CADASIL Family Conference

The CADASIL Association is excited to announce the preliminary phase of planning a 2013 CADASIL Family Conference. The 2013 conference will be held in Boston, Massachusetts on June 27-29. Our Scientific director is Dr. Joseph Arboleda-Velasquez. We plan to have a meet and greet the evening of June 27th, followed by a full conference day on June 28th, and ending with a morning of gatherings to include a CADASIL Association membership meeting on June 29th. All events will be held at the Starr Center at the Schepens Eye Institute (a Harvard Medical School Affiliate). We are working on a group discount for a hotel nearby and hope to keep the registration costs low to allow many in the CADASIL community to attend.

Look for more information on the conference in the February 2013 issue of EMBRACE!

The Starr Center: http://www.schepens.harvard.edu/the-starr-center/core-services/the-starr-center.html

Things to do in Boston: http://www.cityofboston.gov/visitors/thingstodo.asp
Written by Karla: My father passed away from CADASIL one week before my 29th birthday. He was 58 years old. It was not known that he had CADASIL at the time. On his death certificate it stated his cause of death was dementia. We allowed an autopsy to be done, not knowing that we would need that report someday. This report helped to diagnose me. I remember my dad as a very intelligent, strong, stubborn, and hard-working man. My mom, brother, and I watched him getting worse year after year. From what I recall, he started to show signs of CADASIL about ten years before his death. He never had a full stroke, but he had hundreds of mini-strokes. Each time he lost more of his memory, his balance, and other capabilities. We had to put him into a nursing home, and he just got worse and worse. One day the strangest thing happened: I was working a full-time job and something in my head told me to get to the nursing home right away. I told my supervisor that I needed to leave. I went straight to the nursing home and to my dad’s room. He had just passed away, and he was by himself. I was too late! For sure, my dad was extremely loved and is dearly missed.
OU WRITINGS

CADASILIAN’S MEMORY or SOMETHING, I FORGET!
Written by Karla J Smith

Being a CADASILian there are things that we forget to do,
We know to take a shower and give our hair a good shampoo.
We condition our hair and get out of the tub,
When after drying off we realize that our bodies did not get scrubbed.
We decide to start the laundry and think we are doing fine,
When finally we think “Did I remember the detergent this time?”
So off to the dishes all dirty in the sink,
We wash them off and place them in the dishwasher to get clean,
Then we think, “Did I remember the soap?”

After thinking about it we then realize the answer was Nope!!
So we had a busy day showering, laundry and dishes,
It’s time to make some dinner that is going to be delicious.
The smoke alarm goes off and alerts us to the smoky kitchen,
The dinner in the oven was forgotten and now it’s extra crispy chicken.
We decide to throw it out and go get some fast food,
But, the keys are playing hide-and-seek and that is just not good.
So we go through the house looking for the keys,
But then we stop in our tracks and can’t remember what we need!
This is what it’s like to be a CADASILian…

I wrote this poem for you all to read --You are not alone.
Our memory will continue to go and go,
All we can do is laugh at ourselves when these things ensue,
And share our funny stories with all of you.

Setting: Summer 2012

Janet speaking to her husband Andy (as she looks around the living room): Honey, I really meant to mow today, but I'll do it tomorrow.

Andy: You don’t need to mow, baby, I’ll get to it.

Janet (eyebrows raised, perplexed expression): Mow?

Andy: Yeah, you said you meant to mow, but I can do it this weekend.

Janet (laughing at herself): Oh! I meant VACUUM! I was going to vacuum today, but I didn’t.

Andy (scanning the living room carpet): You can mow in here if you think it needs it.

Janet: Excuse me, I guess I just had another brain fart!

(Both laughing)
Please join the CADASIL Association! Membership cards with information about CADASIL and the association will be mailed to every member in 2013. Here’s the link again for your convenience: [www.cadasilassociation.org/Join_Us_2.html](http://www.cadasilassociation.org/Join_Us_2.html)

Looking for ideas on how to help raise money for CADASIL Association toward our goals and mission? Here are some suggestions. We welcome yours, so please share them with us and we will publish them in an upcoming newsletter.

**Gifts:** Looking for the perfect gift for someone on your list? Making a donation to CADASIL Association in honor of someone touched by this disease is a loving gesture.

**GoodSearch.com:** Join GoodSearch to raise funds for CADASIL Association! GoodSearch donates money to your favorite cause when you search the Internet, shop online or dine out at local restaurants! Use GoodSearch.com to search the Internet and they donate a penny per search to your cause. Use GoodShop.com when you shop online and they donate a percentage of every purchase and offer over 100,000 coupons to help you save money too! Sign up for their GoodDining program and they’ll donate a percentage of your restaurant bill when you eat at any one of thousands of participating restaurants. It’s really easy; it’s FREE and turns simple everyday actions into a way to make the world a better place. Please sign up today to help support CADASIL Association! Go to [www.goodsearch.com/nonprofit/cadasil-association.aspx](http://www.goodsearch.com/nonprofit/cadasil-association.aspx) to get started.

**Make Change for Charity:** Toss that spare change from your pocket or purse into a container. Once every few months, locate a Coinstar machine at your local grocery or discount store and donate the cash you receive back to CADASIL Association. This would work well in a school setting as well.

**One Thing I can Do:** Your days are busy, and if you have CADASIL, sometimes filled with painful symptoms. Instead of thinking of all the many ways you can help make a difference, pick just one and follow through on it. One person doing one thing can help toward awareness and ultimately funding toward a cure.

**Know someone who owns a business?** Make a call today and ask if the business could host an event to benefit CADASIL Association. Many businesses in our communities are willing to partner with a good cause. One example: An ice cream shop allowed a charity to display their sign next to a receipt fishbowl by the cash register for one day. Patrons “donated” their receipt. The charity was given 5% of total receipts for the day! Another example: A friend who sells products through home parties donated a percentage of sales at a party organized by a member of the charity. Make that call today!

**Garage sale:** After the holiday season, clean out those closets and pick a date for a spring garage sale! Enlist friends and neighbors who may want to help, and have fun together while supporting CADASIL Association.

Electronic donations are always welcome and greatly appreciated at: [www.cadasilassociation.org/Donate.html](http://www.cadasilassociation.org/Donate.html)

Physical address for donations or other written correspondence:

57 Morgan Place, East Brunswick, NJ 08816
THANK YOU FOR READING OUR NEWSLETTER -- WE HOPE YOU ENJOYED IT!

EMBRACE is created and edited by Karla J. Smith and Janet Mills for the

CADASIL ASSOCIATION

We welcome your submissions of personal stories, questions, news, events, doctor profiles (with permission from the doctor), suggestions for fundraisers, funny or touching creative work, and memorials. We would also love to hear from you with feedback about this newsletter and its contents.

Please email us at: embracenewsletter@gmail.com

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