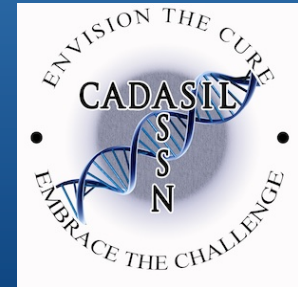


EMBRACE

A newsletter for anyone touched by the rare genetic disease CADASIL

The mission of the CADASIL Association is to raise awareness of CADASIL, ensuring it will be universally recognized and understood by the medical community, enabling patients to be correctly diagnosed. We are dedicated to helping patients, families, caregivers, and other supporters touched by CADASIL. We aim to unite patients and the medical community toward the common goal of treatments and ultimately a cure for this rare genetic disease, by promoting Communication, Advocacy, Research, and Education.



CADASIL Association celebrated one year since incorporation on April 24th!



Join us in this important journey by becoming a member (free of charge) and by volunteering your time to help realize our mission.

Email us at info@cadasilassociation.org

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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: embrace@cadasilassociation.org

OUR STORIES



Emma Chapman
Cairns, Queensland, Australia
Age 37 at diagnosis of CADASIL

I was diagnosed with CADASIL three years ago when I was 37 years old, after suffering a TIA at work. I had always known something was not quite right after my first complex migraine at age 16. I had trouble with numbers, and my concentration and memory were sometimes terrible. (Still are!)

My mum has advanced CADASIL, and both my uncle and grandfather have died from it. I think my younger brother has it too, as he had his first complex migraine a couple of years ago, but he doesn't want to be tested. I have had a few appointments with doctors and neurological "specialists," but they admitted to knowing nothing about this disease, so I self manage now. I take Vitamins B12 and D, Thyroxine and Iodine for my thyroid problem (I believe this disease has affected my pituitary gland, as I suffer with a lot of hormonal problems and sympathetic/parasympathetic nervous system problems), fish oil for depression, and I have cut out processed foods, carbs, and sugars from my diet. I also drink at least 2 liters of water a day. If I do not eat and drink this way I find it affects my health massively, causing headaches, tiredness, and confusion.

I had a nervous breakdown when I was diagnosed, and it has taken two years to come to terms with this. But knowing my life will be different than how I imagined it would be a few years ago has made me grab my life by the throat, live every day, plan for my future, love my friends and family, and never ever take anything -- especially my health -- for granted. It has given me the drive to develop my own business and sort my life out, rather than partying my life away and throwing my money at rubbish, as I was doing a couple of years ago. Knowing I have this has changed my life more than I ever thought it would, but I'm not going to let it ruin it just yet!

OUR STORIES



Jax Tourettes Adams
Weston-super-Mare, England, UK
Age 40 at diagnosis of CADASIL

I was officially diagnosed with CADASIL on Oct 10th, 2012. I am 40 years old and show no symptoms of the disease at all. My mum is 60 and was diagnosed with CADASIL two years ago after lots of guess diagnoses from doctors. My mum has had four strokes and several (unconfirmed) TIAs. She has suffered with migraines, numbness, severe depression, and mood disorders since I can remember. My granddad died at aged 64 from a massive stroke after suffering several TIAs and having Alzheimer's. We now believe he had CADASIL but we can't prove this. He suffered with migraines and depression but didn't talk about it.

After my mum's diagnosis (which she received via letter due to an administration error), my sister had the genetic blood test, as she has always suffered migraines. She has two kids so wanted to test. She got her results last year in person and tested negative. I originally decided not to test as I have no children and never suffered any symptoms. Then I decided I wanted to find out, as I was getting stressed thinking about it. I had genetic counseling beforehand (I was told by my counselor that I had good knowledge of the disease and could go right ahead and have the test done). It took six weeks for the results to come back, and I had to attend the clinic for results in person.

I had decided I would give up smoking on "results day" no matter what. Needless to say, I tested positive for the Notch 3 gene abnormality and I have CADASIL. I asked to be referred to a specialist so I can find out more in-depth about the disease, as I feel different from others because I have never shown any symptoms (as yet).

I feel like I'm living with a time bomb inside me not sure when or if it's going to go off. I did give up smoking on the 10th of October and am still not smoking. I've started to take 75 mg of aspirin daily (my GP "Googled" CADASIL and told me start taking it). I'm doing all I can to lower my risk of stroke. I have lower than average blood pressure, low cholesterol, and I have lost 1-1/2 stone this past year.

I am still coming to terms with my diagnosis and feel like I'm going through a stage of bereavement. My mum says she knows how I feel, but it's hard to talk to her about it as I get upset seeing her going slowly downhill and her memory fading. I don't wear bracelets so I have a CADASIL medic alert tattooed on my inner wrist/arm to alert medics not to give me thrombolysing drugs in case of stroke. I'm told with CADASIL that certain blood-thinning drugs can make situation worse.



Jax's tattoo

ASK DR. JOE ARBOLEDA



Read more about him at:

<http://www.schepens.harvard.edu/arboleda>

Q: Can you tell me about the benefits and safety of Botox for migraines?

A: The FDA has approved Botox for the treatment of chronic migraine. This approval was issued because the benefits outweighed the risks in clinical trials. However, there is no significant evidence that Botox is beneficial or indicated for CADASIL patients. Therefore, decisions of taking this drug should be made in consultation with your primary care physician or neurologists.

Jennette asks: Is it common for patients to get muscle spasms? I have been getting constant pain in my neck and very bad pains in my legs and back.

A: There are no studies linking muscle spasms to CADASIL. However, you should discuss with your doctor a management plan to control this problem.

Maria asks: Does CADASIL affect the healing ability of other health issues? I just had my gallbladder taken out but for months before that I thought it was something from CADASIL.

A: The full spectrum of CADASIL-associated symptoms has yet to be defined. To date there is no evidence indicating an association between CADASIL and healing defects or gallbladder problems. However, all this information is useful for physicians and scientists so I recommend that you discuss these issues with your neurologists.

Send your questions for Dr. Joe to: embrace@cadasilassociation.org and we will feature them in an upcoming issue!

FUNDRAISERS

Opportunities for you to help the Association!

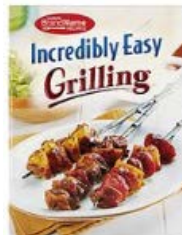
Celebrating Home Online “Party” from May 22nd – July 5th



50% of all sales go to CADASIL Association to support our mission statement.

Click on the link below for access to available items in Celebrating Home’s fundraising program!

www.celebratinghome.com/fundraiser/cadasil/OnlineFundraiserHome.aspx



Inspiring Denim Headbands

\$1 from each sale goes to CADASIL Association to support our mission statement.



These reversible headbands are made from up-cycled denim and cotton fabric covered with encouraging words of strength and hope.

Cost: \$5 each plus shipping

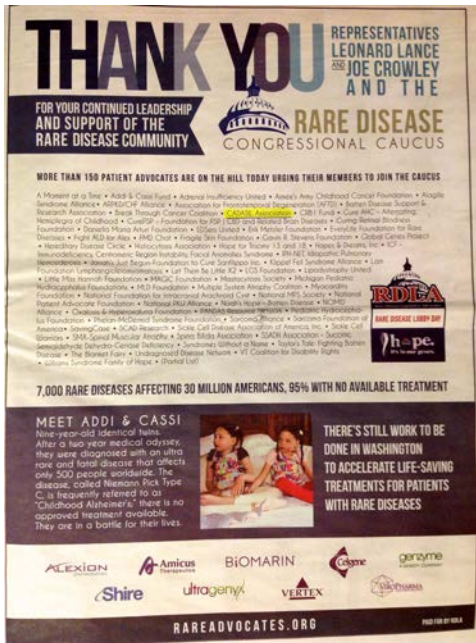
Click on the Etsy shop link below to view and to purchase.

www.etsy.com/listing/150026476/reversible-denim-headband

Thank you for your support!



RECENT NEWS



On February 27, 2013, two political newspapers in Washington, DC published RDLA's (Rare Disease Legislative Advocates) full-page announcement thanking Representatives Leonard Lance and Joe Crowley, and the Rare Disease Congressional Congress, for their continued leadership and support of the Rare Disease Community. The page listed the organizations with patient advocates participating in the 2013 Rare Disease Legislative Lobby Day on Capitol Hill. CADASIL Association is proud to have been represented by four members and recognized on the full-page announcement.

Newspapers *Roll Call* and *Politico* displayed the full-page announcement

On April 9th, 2013, Shire released the "Rare Disease Impact Report," a 34-page document available for download in PDF format at: www.rarediseaseimpact.com (about midway down the page). Association members Janet Mills and her sister Robin are shown on page 9, with a caption that identifies them as having CADASIL. Another step for awareness!



The Mills sisters, both diagnosed with CADASIL syndrome

On April 2nd, 2013, President Obama announced the Brain Initiative with goals that include providing the knowledge for addressing debilitating diseases and conditions.

Read more at: www.whitehouse.gov/infographics/brain-initiative

The February-March 2013 issue of *Neurology Now* features several stories of interest to the CADASIL community. Check this link to learn more: journals.lww.com/neurologynow/toc/2013/09010

Look at articles titled "Young Adult Stroke: Age is Just a Number" and "Vascular Cognitive Impairment: The Brain Needs Blood."

UPCOMING EVENTS

2013 CADASIL Family Conference in Boston

The CADASIL Association is excited to host the 2013 CADASIL Family Conference to be held in Boston, Massachusetts on June 27-29. Our Scientific director is Dr. Joseph Arboleda-Velasquez.

Guest speakers include: Dr. Joseph Arboleda-Velasquez (Schepens/Harvard), Dr. Anand Viswanathan (Mass General), Dr. Stephen Salloway (Brown), Dr. Dev Batish (Athena Diagnostics – which is the genetic testing lab for CADASIL in the USA), Dr. Adrian Ivinson, (Harvard Neurodiscovery), Sigurros Davidsdottir, (Mass General

All sessions of the conference will be held at the Constitution Inn. Call the hotel at 617-241-8400 or 1-800-495-9622 to reserve your room for \$125 a night when you mention the CADASIL Conference. You must reserve your room by May 27th to receive the discount.

The schedule includes:

Thursday June 27, 2013 - Meet & Greet with catered finger foods from 7:00pm - 10:00pm at the Constitution Inn. Please sign in during the Meet & Greet. If you cannot attend, you may also sign in the morning of the conference.

Friday June 28, 2013 - Continental Breakfast from 8:00am - 9:00am at the Constitution Inn. Conference from 9:00am-5:00 pm with a catered lunch from 12:30pm - 1:30pm.

A large part of the conference will be a Question and Answer session, so have your questions ready!

Saturday June 29, 2013 - Continental Breakfast from 8:00am - 9:00am at the Constitution Inn followed by a CADASIL Association Meeting from 9:00am - 10:00am with breakout sessions for patients, caregivers, and those undiagnosed who may be trying to decide if they want to be tested. ALL INVITED! If you are not already a member of the CADASIL Association, please sign up on our [Join Us](#) page on this website, or you may join at the conference. Membership is free.

Registration fees and more details can be found at: www.cadasilassociation.org/2013-boston-conference

Things to do in Boston: www.cityofboston.gov/visitors/thingstodo.asp



Our thanks to Karl Stumpf for the design of the Boston Conference logo!



MORE UPCOMING EVENTS

The Ride for CADASIL Fundraiser in St. Paul

When: Saturday, June 8th, 2013

Where: Motorcycle Ride @ Hitching Post Motorsports, So. St. Paul, MN www.hponline.com

Register: 9:00-10:00 a.m. Ride Departs: 10:30 a.m.

Events:

- Motorcycle Ride
- Silent Auction: 5:00 -7:00 p.m.
- Dogs & Brats: 5:00 - 7:00 p.m.
- Entertainment: 5:00 - 9:00 p.m.

Coordinator/Contact: Bob Leslie (612) 801-0336, WingBob2005@msn.com

Web Site: www.cadasilrally.org

Facebook: Ride for CADASIL Fundraiser

Proceeds go to Butler Hospital for CADASIL Research



Check out page 12 of this newsletter for ways you can participate to help the global CADASIL community!

OUR WRITINGS

The Stigma of Rare Disease: How Do You Tell People About Your Condition? by Janet Mills

Posted on April 17, 2013 at: globalgenes.org/the-stigma-of-rare-disease-how-do-you-tell-people-about-your-condition

Many rare diseases, like my own, have hard-to-pronounce names that evoke fear or boredom from people first hearing them. In my experience, eyes glaze over around the middle of “Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy.” Giving the acronym “CADASIL” for short doesn’t help much either, as most people have no clue what it is.

While I imagine a majority of us in the rare disease community have studied the science behind the big words of our conditions and have reasonable ideas of what they mean, the names of our diseases can sound like a foreign language to the average person. How do we get around that and explain what’s happening to us? I believe that bringing the words down to understandable levels can help avoid some of the stigma of having a rare disease. Here are some ideas I have used:

- **If the disease is not contagious, say so.** It is natural to shy away from someone who could make you sick. I sometimes joke, “I promise not to rub my brain on you.” Of course, no one can get CADASIL from touching my brain, but saying something to lighten the moment and assure the person of her safety can help. If the disease *is* contagious, giving information up front about how it is transmitted can relieve some anxiety.
- **Keep it simple.** Consider naming a less-rare condition that most people know and relating it to your disease. One of my lines is, “CADASIL is often misdiagnosed as MS (Multiple Sclerosis) because our brain images and some of our symptoms are similar.” I then go on to tell them how CADASIL is different from MS and share the importance of knowing which condition a person has. In the world of rare cancers, simply saying the word “cancer” will provide immediate understanding at a basic level. Then you can give more details about the specific type.
- **Tell how the disease affects you.** From your appearance and physical condition, it may be fairly clear what the disease is doing to you. Chances are, however, that a lot more is going on than what people can see. Some rare diseases cause “invisible disabilities” and, to someone unfamiliar with the disease, the patient looks perfectly fine. CADASIL is one of those. A CADASILian who has not yet had a stroke may not show any outward signs of the disease. If people could view our brain MRIs and get a glimpse of our compromised blood vessels, they would realize the seriousness of the condition. Instead of carrying around medical records and images, explaining how it feels to have your condition is helpful to others. When people tell me “You look great! Are you feeling better?” I tend to respond with something like “Thanks. Today is not a Couch Day. You won’t see me out and about on those days.” A reply like this is polite and subtle, reminding the person you are still sick.
- **Carry cards about your disease.** Online printers such as Vista Print (www.vistaprint.com) offer custom business cards for a low cost. A small image along with a brief explanation of the disease and a list of symptoms can fit on one side. I keep about a dozen CADASIL cards in my purse. They display information about the disease as well as contact details for CADASIL Association (www.cadasilassociation.org), so someone interested can find out more. At first I was too emotional and hesitant to hand out these cards. Over time, I have become more assertive and an advocate for my rare disease. Anytime I am asked where I work, I pull out a card to help explain why I don’t. I give the cards to friends who can’t remember what my disease is called. I offer them to strangers I’ve just met at a social gathering when they ask what I do, or to someone with whom I strike up a conversation.

There are many things we can do to relieve the stigma associated with rare diseases. The ideas here are just a sampling. The public is becoming more aware of us as a group, thanks to the efforts of Global Genes (www.globalgenes.org) and other active organizations. We can help in our own ways by simply sharing our stories.

PARTICIPATE

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

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 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

Physical address for donations or other written correspondence:

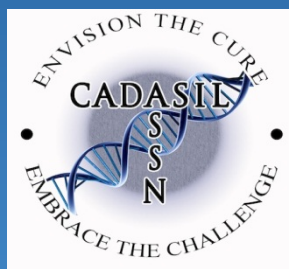
P. O. Box 124, Helmetta, NJ 08828

THANK YOU FOR READING OUR NEWSLETTER -- WE HOPE YOU ENJOYED IT!

**EMBRACE is created and edited by Janet Mills and Karla J. Smith 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: embrace@cadasilassociation.org



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CADASIL Association

P. O. Box 124

Helmetta, NJ 08828

Website: www.cadasilassociation.org

Email (general): info@cadasilassociation.org

Email for Embrace: embrace@cadasilassociation.org

To leave a telephone message: 307-215-9840