

EMBRACE

A newsletter for anyone touched by the rare genetic disease CADASIL

The mission of CureCADASIL/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.



Please remember cureCADASIL in your end-of-year or New Year gift giving. You can help fund vital research!

Donations may be made via check or PayPal.

cadasilassociation.org/donate

December 2015 - Issue 8 Part 1

Too much news for just one 2015 issue! Be sure to see Part 2!

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Look what we're up to!

We have more to share!
**Be sure to read our 2015
Part 2 Issue!**

Please join our cureCADASIL Family Registry!

We need:

*Diagnosed CADASIL patients

*At-Risk family members

*Deceased patient and/or suspected patient info

cadasilassociation.org/curecadasil-registry

Why should my family join the cureCADASIL Family Registry?

 Because you can make a difference in finding a cure.

Want to tell your story? Have a question for the doctor? Planning an event? Upcoming newsletters 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, memorials, and events. Please email them to: info@cadasilassociation.org

OUR STORIES



Laura Luhrsen
Formerly of Redlands, California
Now living in New York
Diagnosed at age 51

I try to stay positive, but it's hard since I've recently been diagnosed with a rare genetic terminal illness that has no cure! I suffered a stroke on 9/20/13, but I was not diagnosed with CADASIL until the summer of 2015. I've had five major surgeries since my stroke, which occurred one week after my wedding, on my honeymoon that never happened at the river in Laughlin, Nevada.

I spent the next nine days in the ICU in Las Vegas, the closest neurological hospital near Bull Head City. The next five weeks I was an in-patient at Loma Linda University Medical Center, tied to a chair with no core, loss of speech, loss of mobility, everything! My brain was still there, but my body was foreign. It still is.

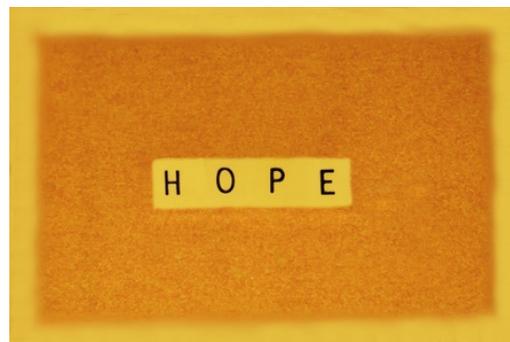
I guess I'm blessed that I did know how to play the piano. I did hike and ride a bike. I did have three beautiful daughters who have contributed wonderfully to society. I did have a chance to care for my mom for nine years.

I want it all back, but I'm trying to be humble now and realize there are others who are less fortunate than I am. I will count my blessings and move on...

I can deal with the reason I had my stroke and that my mother was misdiagnosed. She must have had CADASIL as well. But I'm having a really hard time knowing that my three daughters have a 50/50 chance of having this autosomal dominant gene.

There's nothing I am able to do but pray! GOD is the ULTIMATE PHYSICIAN... My former LLUMC neurologist "let me go" as there is no cure.

I'm helping bring awareness to this awful mutation of the Notch 3 gene on the 19th chromosome in the DNA strand.



OUR STORIES



Donna Martin Roswell, New Mexico Husband Mike diagnosed at age 57

My husband, Mike, comes from a long line of CADASIL patients. His grandfather had it, or died of a series of strokes. He was not really diagnosed back then. Mike remembers his father having had strokes. His father would get so angry when trying to speak and express himself that he eventually just quit talking. Mike was only 11 when he lost his father. His father, also, was not diagnosed with CADASIL, but due to the strokes and passing away from stroke related conditions, it was pretty much a given when the children began having the same symptoms and being diagnosed.

Modern medicine connected the dots. Mike was the youngest of 11 children: 9 boys and 2 girls. One boy died accidentally, so it is unknown if he carried the mutation or not. All surviving boys had CADASIL. Two of the boys were twins. As time progressed, the family learned of the Familial Stroke Syndrome and how it might relate to them. In 1997, what was left of the family went to Mayo Clinic in Rochester, Minnesota, as they were doing a study on CADASIL and sponsored different families for the research. All boys, except Mike, were presenting symptoms. Mike was not tested, but the diagnosis was made for most of the other participants. Oddly, they mostly displayed different symptoms. One would cry for all emotions, happy, sad, mad, glad, or whatever the event. He would cry, even when it was a happy occasion. Another brother would often laugh uncontrollably. That is just how CADASIL presented in them. They just could not control the laughing or crying. The same brother that would laugh was also losing his ability for speech. At first it was just difficult to understand him. After several more strokes we could not understand him at all. He was functional otherwise. The twins did not present in a similar way at all. One twin went immediately to the nursing home. He needed more professional care than could be given him at home. The other twin lived considerably longer, and slowly developed dementia over many years. One brother began having strokes at 23, and he is 8 years older than Mike (Mike is now 62). He has experienced a sort of fainting spell or light seizures. He had a pretty big stroke, but recovered most functions after a long bout with therapy. He is living at home and, for the most part, doing pretty well. Each of the remaining boys also display different symptoms, probably because of the strokes hitting in different regions of the brain, AND the differences in personality of each brother.

Now, there is only Mike, one brother, and one sister living. Neither of the two girls showed signs of CADASIL, but one died of breast cancer. Mike had said several times, "I don't know how long I am going to be able to outrun this." He was the only one who did not show any signs of CADASIL. Then in January of 2008, we lost the race. Mike had a major stroke. He lost use of his right arm and leg and speech. During the intense therapy in the coming months he was able to regain use of his leg and most of his speech. His right arm was still not functional. Since he was right handed, this created yet another problem. But, he could walk and talk pretty well. We practiced a lot at home and more of his speech returned. Eventually he was able to walk on his own without the cane. There was still a limp. We had to make a lot of adjustments on how we did things, but he was functional again. He could drive, work, walk, sit, stand, dress...just slower. In 2010, we went to Mayo Clinic again and Mike was tested positive. It was no shock or surprise, due to it having been all around him all of his life, and having had the stroke.

Donna & Mike Martin, continued

After Mike had his first stroke in 2008, he had about an 80% recovery. Our life had pretty much resumed close to normal. Then he broke his hip in August 2013. He had emergency surgery for partial hip replacement. He has never recovered from the surgery. Had the surgeon researched CADASIL a little further to see if there is a favored anesthesia for CADASIL, perhaps the outcome would be different.

A number of months ago I noticed a gradual mental decline. I began a dreadful journey within myself, discovering feelings and emotions that I never thought could ever be part of me. It is a living nightmare that continues on and on, slowly getting worse. I'm against the wall, nowhere to turn, no help in sight. I am all we have. I am bombarded from all directions, often overwhelmed, and I don't know what to do.

Most of us, non-professional caregivers, are simply spouses or family members. We have had no formal training or experience of any kind in this field. Overnight we are thrust into a whole new career that we know nothing about, and we may or may not be able to handle it. Plus, we are generally *held accountable* for a job we know nothing about, and the welfare of our patient is on us. If the patient gets better, the physician or therapist gets the thumbs up. If the patient gets worse or does not improve, it must be the care they are getting at home. And the nightmare marches on.

Sometimes when frustrated, Mike is able to speak clearly, and when he can it is cuss words or hateful, ugly things that are so terribly uncharacteristic for him. My heart breaks when I see the tears in his eyes when he cannot say what he wants to and the frustration is more than he can bear. With each month he is able to talk less and less with any continuity. He cannot carry on a simple conversation and is distracted very easily. I try to talk to him, but he just stares at the TV. I feel ignored, yet I know he can't help it. But, it still hurts. The little hits keep coming and the hurt just grows and eats away at my heart, creating an opening for unintended resentment. I cannot include him in any of the decision-making. He just is not mentally capable of even making simple decisions. He cannot stand or walk unassisted. I have to transfer him from wheelchair to bed or chair or whatever. Sometimes he works with me and sometimes he seems to just do what is in his head. He gets angry, and justifiably so, but then that puts his safety in jeopardy. I get upset, then angry, and it shows, or I mess up and say something less than kind back to him. I see the hurt in his eyes. He knows he is hurting me and does not mean to.

It is just the two of us. There is no help unless we have an emergency. No help on a day-to-day basis. I had to quit a job of 28 years that I loved. It was high traffic, high interaction, and a lot of contact with a lot of people who had become friends. But I loved Mike more than the job, and I just could not let a stranger take care of him. The last day on the job was a terrible day for me. And I had to do it alone. When I came home, a crying mess, he didn't even notice anything was wrong. He was no longer able to understand things like this, where before he was the rock I ran to and my fixer. He was big enough and strong enough to slay all of my dragons. But, now that man is gone, and I truly miss him. When I tell him something is wrong, he can't remember it for any length of time. If something happened to me and I was out of commission for a few days or weeks, I do not know what would happen to him. I worry so much about this.

Sometimes I get so angry at him or so hurt by him, that I find sides of myself I never knew were there. Trying to contain this anger and hurt only makes me more volatile. I know I have come close to the edge more than once, which creates more fears and issues. I only sleep a few hours a night due to fear of everything that one can be afraid of. I cannot imagine myself without this man, yet I see the end coming as sure as I sit here.

Donna & Mike Martin, continued

I am generally pretty confident and strong. But Mike is my one big weakness, and seeing him this way with the constant changes for the worse, I feel parts of me slipping away. On a daily basis I have to cope with the high level of stress, fear, anxiety, lack of sleep, isolation, and all of the other deadly emotions that this hideous disease has cursed us with. I watch little pieces of him drift away. I hold tight to whatever thread of hope that is offered me that day, but by nightfall, it is gone. The tiny rays of sunshine are suffocated by the darkness of the recurring nightmare that is actually our reality.

I have not even touched on my feelings of guilt. I have this horrible guilt because I am complaining about what I am dealing with, when I know he is the one with the *real* monster. I cannot imagine how I would react to my being in his place. With what he is going through, how can I have the audacity to complain? All I know is that I do have these feelings and emotions and they are real and running wild. They are part of me and if I do not control them, I will end up inflicting even more misery on him and myself. My attitude will bleed over onto him and his problems will only magnify. Not to mention my sanity becoming confetti in the wind. I can fully understand why there are so many dementia caregivers that are totally dependent on alcohol and/or drugs. They dull the unintended hurt and falsely placed guilt. The physicians do not even think about our wellbeing.

In my opinion, the caregiver is just as important as the physician, maybe even more so because they love and are always there for the patient. The caregiver should be just as important to the physician for they have such a tremendous impact on the patient. The caregiver's mental and emotional state can definitely inject the patient with a negative or positive result in attitude. Caregivers have to live *two* lives. On a 24/7 basis, they have to try to help the patient live their life as comfortable, clean, safe and happy as possible, which is a full time job. In addition, they have to live their own lives, which is another full time job of washing, cooking, cleaning, errands, appointments, groceries, bill paying, keeping up the house, yard, car, etc. Before, it took two people to handle all of these daily chores. Now there is only one.

There is an imperative need for physicians to pay more attention to emotional problems in CADASIL patients. *In addition*, it would help so very much if they paid more attention to the caregiver's emotional issues as well. There should be an information source of some kind that goes hand in hand with the patient *and* caregiver. The physicians should consider both parties involved. If the patient is angry, aggressive and violent, then the caregiver is going to be the recipient of this turmoil and would need different counseling and/or meds than someone who might have a patient who is very passive and does or says nothing. In this case, the caregiver is alone and isolated. These are only two of the countless displays that should be considered by the physicians. *Both* patient and caregiver should have equal time and consideration. Each has such a powerful impact on the other. Sometimes the caregiver is in greater need of mental or emotional help than the patient. Most caregivers are at such a loss with CADASIL, that we are wandering around in the dark grabbing at straws and searching for tiny rays of sunshine. We are either free falling or slammed against the wall.

When the physician requires the "New Patient Form" to be filled out, there should be a second form for the caregiver. The physician should consider the caregiver as their patient too. Giving them advice, information, and possibly medications. The caregivers may need help coping with traumatic changes in their lives and the lives of their loved ones. Physicians can possibly prevent an additional CADASIL casualty.

For every CADASIL patient there is a second patient, the caregiver.

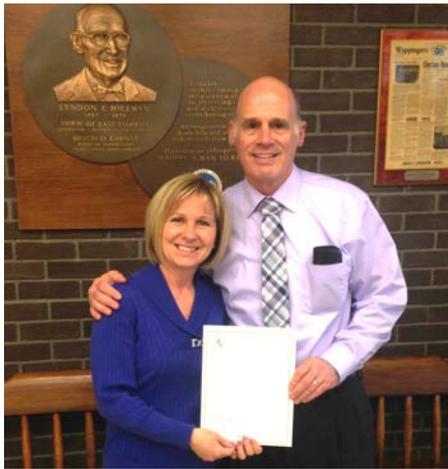
PHOTOS & NEWS



Our volunteers are working for the CURE!

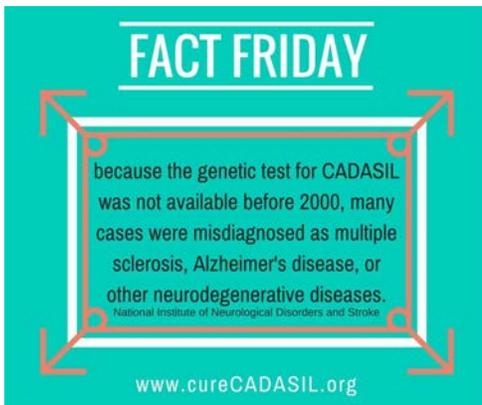
Photos from conferences, meetings, and awareness events attended by cureCADASIL trustees and members in 2015

Please share what you are doing so we can include you in EMBRACE! Email us at: info@cadasilassociation.org



Janice Ragazzo frequently meets with her legislators in Washington, DC and in her home state of New York.

Have you seen our Fact Friday posts from Nancy Maurer on our Facebook and Twitter pages?



If not, please be sure to follow us at:

[facebook.com/CADASIL.Association](https://www.facebook.com/CADASIL.Association)

and twitter.com/CADASIL_Assn

MORE PHOTOS & NEWS

In February, cureCADASIL introduced a new 30-second public service announcement to spread awareness of CADASIL. The video features actor Jim O'Heir from TV's "Parks and Recreation." Please share it and show it to anyone who does not know about CADASIL!

Watch at: youtu.be/SI2WwaR19Gc

cureCADASIL wishes to thank actor Jim O'Heir and "Parks and Recreation" Associate Producer Tom Ragazzo (son of Trustee Janice Ragazzo), for making this production possible!

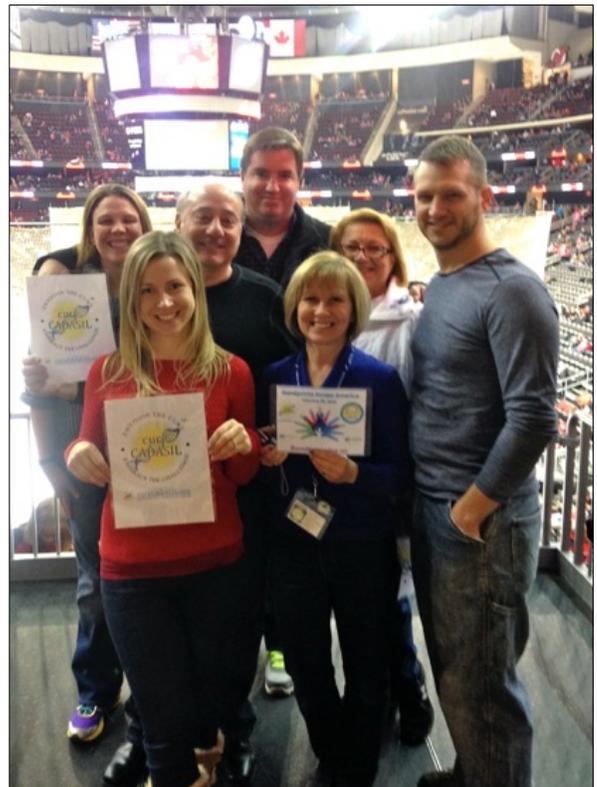


cureCADASIL's Ronnie Bradbury and Anne McGuinness at the New Jersey Rare Disease United Awareness Event at the RWJ (Robert Wood Johnson) Health and Wellness Center in Hamilton Township, NJ, where they educated many medical professionals and others about CADASIL.

Almost \$5,000,00 was raised for cureCADASIL as the recognized non-profit organization at the New Jersey Devils NHL hockey game vs. Arizona in Newark, NJ. We are excited to announce a second opportunity to be highlighted at a New Jersey Devils game against the Philly Flyers on February 16, 2016. For tickets, please contact us at: info@cadasilassociation.org

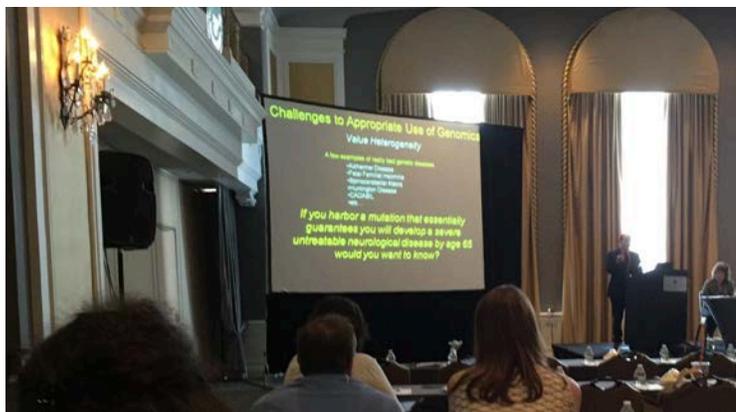
A big thank you goes out to Tony and Janice Ragazzo for their efforts toward this fundraiser!

Pictured from L-R: Michelle McGuinness, Kristen Ragazzo, Tony Ragazzo, Matt Powers, Janice Ragazzo, Anne McGuinness, and Dan Mascaro



MORE PHOTOS & NEWS

Right: cureCADASIL's Barbara Hunt, Janice Ragazzo, and Anne McGuinness attended the AAN/American Academy of Neurology's Annual Meeting in Washington, DC in April 2015.



Above: Anne McGuinness attended the New York-Mid-Atlantic Consortium for Genetics and Newborn Screening Services (NYMAC) Summit 2015 from May 28-29 in Baltimore, MD. NYMAC is one of seven regional collaboratives within the United States that aims to improve access to genetic services for families affected by inherited disorders. Speakers included parents, genetic counselors, geneticists, physicians and other health-care providers, policy makers, and other stakeholders. Topics focused on overcoming barriers that prevent individuals and families from receiving genetic services. Anne set up a table with CADASIL information. She attended a breakout session with the NYMAC Public Health Workgroup and joined this workgroup. Through this NYMAC workgroup, a CADASIL webinar was produced for medical and healthcare professionals. Dr. Swati Sathe and Anne McGuinness participated in this webinar.



The 1st Annual "Soaring Above the Storm" run/walk fundraiser was held May 9th in New Jersey, USA. Our thanks to the McGuinness/Montgomery/Culcasi family for all their efforts to make the event a success! Watch for updates on specific dates and info about the May 2016 event.



This poster was displayed at the May 2015 event.

MORE PHOTOS & NEWS

More photos from The 1st Annual “Soaring Above the Storm” run/walk fundraiser held May 9th in New Jersey, USA.



MORE PHOTOS & NEWS



In July 2015, a group of cureCADASIL trustees, scientific advisors, and members attended the ULF/United Leukodystrophy conference in Omaha, Nebraska. cureCADASIL Scientific Advisor Dr. Swati Sathe, of Rutgers Medical School, spoke about CADASIL. The awardee for the ULF's 2014 CADASIL Research Grant, Dr. Fabrice Dabertrand of the University of Vermont, was also a highlighted speaker for CADASIL. (Many members of cureCADASIL Association contributed to the ULF CADASIL Research Grant Fund over several years.) We were fortunate to have these experts all to ourselves for several hours one evening during the event.

Above from L-R: Janet Mills, Anne McGuinness, Robin Spaur, Dr. Swati Sathe, Dr. Fabrice Dabertrand, Laura Luhrsens, Sue Martin, and Michelle McGuinness.



Right: Patient and advocate Laura Luhrsens chatted with Derek Gavin, the Director of Development at NORD (National Organization for Rare Diseases) in Omaha.



SUMMARIES OF 2015 NEWS

Janice Ragazzo attended Rare Disease Week on Capitol Hill in Washington, DC on February 23–27 as part of the RDLA (Rare Disease Legislative Advocates) group. Read more about her visit below:

On February 25th, Janice Ragazzo met with the legislative assistants from N.Y. Senator Kirsten Gillibrand, N.Y. Rep. Paul Tonko and Rep. Sean Maloney's offices. Janice received a letter of endorsement from Assembly Kieran Michael Lalor to present to Rep. Sean Maloney showing support for cureCADASIL Association's efforts to have CADASIL receive its own ICD code for inclusion of ICD-11 in 2017. Also, upon Janice's request, he urged Congress to support H.R. 292, which would amend the Public Service Act to provide for systematic data collection and analysis and epidemiological research regarding Multiple Sclerosis, Parkinson's Disease, and other neurological diseases. He stated that CADASIL should be added to the list. Janice left cureCADASIL Association packets with each office, which contained vital information on our work on raising awareness for CADASIL. (Janice received a phone call on March 6th from Thomas Mintz, Senior Legislative Assistant to Rep. Sean Maloney in Washington, to let her know that the Representative signed on to RH292.)

On February 26th, Janice Ragazzo attended the Rare Disease Congressional Caucus Briefing, and later in the afternoon NORD's special preview of "Banner on the Moon," a movie about a rare patient's journey. It was here that she met and spoke with David B. Flannery, Medical Director of American College of Medical Genetics and Genomics located in Bethesda, MD. David was part of the international medical team that just finished work on the ICD-10 codes (International Classification of Diseases) due out this October. Since then, Janice has been in touch with David to follow up on having CADASIL classified.

Finally, on February 27th, Janice Ragazzo attended Rare Disease Day at the NIH (National Institute of Health) in Bethesda, MD. There she met with Adam Bennett, in charge of international sales for Gene D (a DNA Diagnostic Company). She noted to him that currently they do not have CADASIL listed as one of the disorders. Since then, Adam has notified Janice that he has passed on all the CADASIL information she gave him to his colleagues on their neuro team and is currently awaiting feedback.

Janice has stated how beneficial it is to be in Washington for these important events that are devoted to raising awareness for rare disorders, especially CADASIL. She strongly suggests that other members from the CADASIL community try to partake in the 2016 session.

In March, cureCADASIL participated in our first GiveRare Day, where donations totaled \$155. Thank you to our supporters!

Anne McGuinness and Janet Mills had a phone conference with Bob Rauner, the President of the ULF (United Leukodystrophy Foundation) in regard to having CADASIL be a highlighted disease during the ULF Annual Conference in Omaha, Nebraska in July 2015. Several cureCADASIL.org trustees and members ultimately attended (see page 10).

Janet Mills was a call-in guest on Gary Garver's "Controlled Chaos" Radio Show in Loma Linda, CA (Los Angeles area). This was Janet's third on-air chat with Gary. She shared more information on CADASIL and how cureCADASIL.org is funding important research. Gary's brother is a CADASIL patient. <http://www.kcaaradio.com>

MORE SUMMARIES OF 2015 NEWS



On April 1st, Janet Mills was a guest panelist for Global Genes' webinar about genetic diseases. Her portion of the discussion included talking about her diagnosis of CADASIL and what symptoms pushed her to seek medical help. She made sure to say that she'd first been diagnosed with MS/Multiple Sclerosis, and shared the steps it took to be diagnosed correctly. This continues to be a problem in our CADASIL community. Janet also talked about how she disclosed the information of her diagnosis to her family and friends. The full webinar, titled **How to Discuss Genetic Disease with Your Loved Ones**, can be viewed here: globalgenes.org/april2015webinar

Anne McGuinness, Ronnie Bradbury, and Janet Mills had a phone conference with Dr. James from Avanir Pharmaceuticals in regards to their prescription medication Nuedexta for the symptom PBA (pseudo bulbar affect) which can cause involuntary, sudden, and frequent episodes of laughing and/or crying. Some CADASIL patients report having PBA. (Please read Donna Martin's story on pages 3-5.) <http://www.avanir.com/nuedexta>

cureCADASIL signed the letter in support of the Rare Pediatric Disease Priority Review Voucher (PRV) Program.

cureCADASIL/CADASIL Association Trustees met with Athena Diagnostics executives at the American Academy of Neurology Annual Meeting in Washington DC. We negotiated with Athena Executives to provide CADASIL patients with the resources they need to obtain genetic testing for CADASIL and educational materials to help them interpret their results. The Athena Alliance program is a new program based on income levels. Please see the Income Eligibility Chart in the link below. For the lower income level, a patient will never have to pay more than \$250.00. For patients who income falls in the upper income brackets, they will pay \$600.00 to \$800.00, depending on their income and family size. cureCADASIL/CADASIL Association Board members will continue to meet with Athena executives as needed. <http://www.athenadiagnostics.com/insurance/athena-alliance-program>

On April 27th, cureCADASIL/CADASIL Association Inc. held its General Membership and Public Annual Meeting via conference call to inform members and anyone else who is interested of our recent accomplishments, activities, and upcoming goals.

Linz Culcasi was interviewed by Gary Garver of KCAA Radio about her "Soaring Above the Storm" event. KCAA radio is based in California and their livestream and podcasts can be heard by anyone through links at the website. <http://www.kcaaradio.com> (See event photos on page 9.)

Linz was also interviewed by Burt Baron. <http://www.wctcam.com/BertPodcasts.aspx>

DON'T MISS PART 2 OF OUR 2015 EMBRACE NEWSLETTER

which includes:

Updates on CADASIL Research
(funded with *your* donations)

RDR (Rare Disease Report) News

More Event News

Upcoming Events

Another great article written by Phil Jones

Meet-Ups

Ways you can help cureCADASIL

And more!

THANK YOU FOR READING OUR NEWSLETTER!

EMBRACE is created and edited by members of cureCADASIL 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: info@cadasilassociation.org



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