Deater Foundation, Inc.
PO Box 255
White Deer, PA 17887

Deater Foundation Newsletter 2021

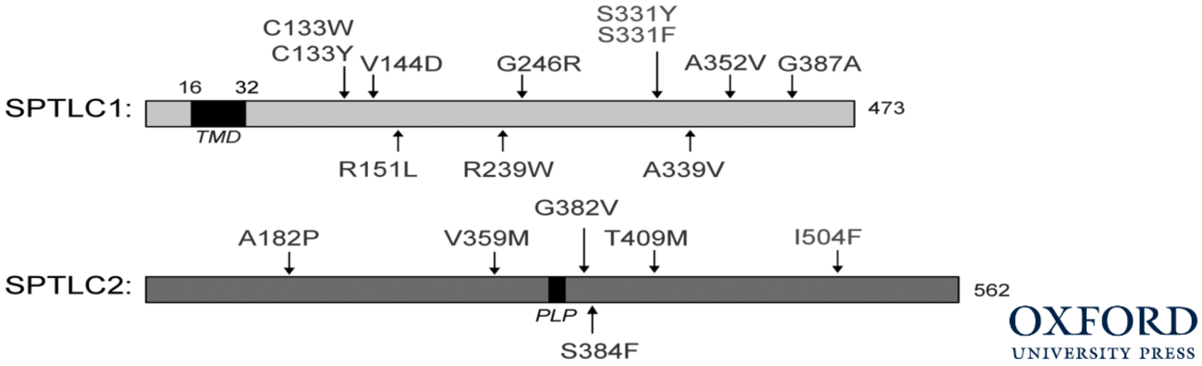
The purpose of the Deater Foundation is to provide funding for medical research for the disease Hereditary Sensory and Autonomic Neuropathy Type1 (HSAN1) to discover a treatment or cure.

HSAN1 is a rare inherited peripheral neuropathy. Each child of an affected parent has a 50 percent chance of inheriting the affected gene. HSAN1 patients develop severe loss of sensation to temperature, pressure, and pain. The loss starts in the extremities, usually beginning in the feet. This sensory loss often leads to painless injuries, chronic skin ulcers, bone destruction, bone infections, pain, amputation of fingers and toes, and, eventually, difficulties with movement.

HSAN1 symptoms typically begin in the late teens. The diagnosis is made by genetic testing. HSAN1 is progressively debilitating and severely disabling and should be treated urgently. Many affected patients often notice weakness and muscle atrophy, and require surgeries or walking aids such as canes, braces, or wheelchairs.

HSAN1 is caused by mutations in the genes *SPTLC1* and *SPTLC2*. These genes encode the serine palmitoyltransferase (SPT) enzyme. SPT controls the production of sphingolipids, a fat important for normal neuron function. In HSAN1, SPTLC mutations alter SPT so that the enzyme produces abnormal fats called deoxysphingoid bases (DSBs). These accumulate in sensory neurons, impairing the cells' proper function and leading to HSAN1 symptoms.

www.neuropathycommons.org/genetics



OXFORD UNIVERSITY PRESS

The Deater family has experienced this disease for at least seven generations. Members have participated in research regarding the cause, treatment, and potential cure since 1939. In 1990 members of the family established the Deater Foundation, Inc. (DFI) to raise funds for research and to increase awareness of this debilitating disease.

Dr. Robert H. Brown, Jr. University of Massachusetts Medical Center

At the University of Massachusetts (UMass) Medical Center, Dr. Bob Brown reports that he and a graduate student, Huiya Yang, are collaborating with two teams to generate a new line of mice that harbor the specific mutation in the mouse SPTLC1 gene corresponding to the Deater HSAN gene mutation, C133Y. The team, with Drs. Florian Eichler and Alex McCampbell, previously reported producing a line of mice that developed HSAN1 because they had several extra copies of the SPTLC1 gene with the HSAN1 mutation. If all goes well, the new mice will have the Deater mutation embedded in the mouse gene itself. This differs from the previous models we have published in which we leave the mouse SPTLC1 genes intact but add external, so-called transgenes that have mutant SPTLC1.

The goal is to have a mouse model that exactly reproduces the Deater form of HSAN1. This, in turn, should be immensely valuable in studies of therapies. Toward that end, the UMass team, aided enormously by Dr. Jonathan Watts, a faculty member in the RNA biology unit at UMass Medical, is now also generating new molecular tools (anti-sense oligonucleotides or ASO) to suppress the mutant gene, first in Deater fibroblasts and then in the mice. Anti-sense oligonucleotides are synthetic short DNA or RNA molecules that can interact so that complexes called hybrids are formed by molecules with similar, complementary sequences. They have been successfully employed to inhibit gene expression in patients with other neuromuscular disorders.

So far the ASOs look promising in the fibroblast cell culture studies.

Deater Foundation Inc. and Dr. Oaklander/ Neuropathy Commons Collaboration

Tami Newcomer Murphy

Beginning in the spring of 2020, a few members of DFI met virtually with Dr. Anne Louise Oaklander and her team. Dr. Oaklander is the Director of the Nerve Unit and Neurodiagnostic Skin-Biopsy Laboratory at Massachusetts General Hospital. She specializes in peripheral nerve disorders and small-fiber polyneuropathy, in adults and children. The purpose of these meetings was to discuss areas of her team's work and how DFI might help. During these virtual meetings, Dr. Oaklander's team shared their aims and goals, and DFI shared with the group about our history and mission as well.

The first area involves an upcoming NIH R01 grant that is focused entirely on pediatric neuropathy, including genetic testing. This grant could help create and optimize standardized, validated metrics so people (patients, doctors, and researchers) are all speaking the same language and using the same tools for testing when it comes to HSAN1. DFI could offer support for this work in terms of outreach and education, and possibly with patient testing/tracking.

Second, is the need to create a centralized HSAN1 Patient Registry. This registry would be housed in a secure online location and would be a tool for data collection, repository, and analysis. Both HSAN1 patients and their doctors would have access to this registry to allow for an increased standard of care. Researchers would also have access to this registry to advance disease understanding and development of potential treatments.

The third area focused on ways to increase HSAN1 visibility and information through the MGH platform, NeuropathyCommons. The Executive Editor of this website, Patricia McCaffrey, has been helping to coordinate the sharing of information/links between http://www.deaterfoundation.org/ and their site https://neuropathycommons.org/ . NeuropathyCommons has included a page for HSAN1on their website. It draws extensively from the DFI website with links back to DFI. Our mutual hope for this collaboration is to bring about increased awareness and information about HSAN1.

Interview with Ginger Satterfield Scally

Ginger Satterfield Scally is a student at the Southwest College of Naturopathic Medicine in Tempe, Arizona. She is in a four-year program to become a Naturopathic physician. Because of several factors, including her diagnosis of HSAN1 she has decided to take six years to complete the program. Ginger is using her medical knowledge and personal experience to advance the cause of treatment for HSAN1.

Ginger's mother, Fran Satterfield, died eight years ago at age 70. She had extensive neuropathy but was never diagnosed with HSAN1. Ginger had the same symptoms as her mother and suspected that there might be a genetic component to their issues as they were nearly identical in manifestation. When her mother passed away from an accidental overdose of her pain medicine, Ginger decided there had to be a better way and that set her on the quest to discover naturopathic medicine.

Ginger began to experience sensory problems in the seventh grade. She encountered a medical system that attributed her symptoms to being "all in your head". She was "diagnosed" with peripheral neuropathy with no known etiology (numbness or pain usually in the extremities arising spontaneously with no known cause). In 2015, while living in Michigan, she was sent to a neurologist who acknowledged that he suspected her neuropathy had a genetic component but refused to do testing and actually made the comment, "We know it is genetic because you have exactly what your mother had. Even if we tested you, there is nothing we could do about it." She did not give up.

After moving to Arizona for medical school in 2017, her naturopathic doctor referred her to a neurologist who respected the way naturopaths do things, with as little pharmaceutical intervention as possible. Ginger was adamant about having genetic testing. The neurologist also ordered electromyographic testing, a spinal tap, nerve conduction tests and a sural nerve biopsy. All tests showed significant nerve damage. It took six weeks to get the results of the genetic tests back. Finally, a diagnosis! She was told she had Charcot Marie Tooth, but the doctor had downloaded the wrong disease information. When Ginger arrived home with the genetic report, she was able to see that she actually has Hereditary Sensory and Autonomic Neuropathy Type 1A with a V144D mutation.

Ginger found the HSAN1 Facebook support group and subsequently the www.deaterfoundation.org website. She reached out to various researchers and found Thorsten Hornemann, PhD at the Institute of Clinical Chemistry, University of Zurich, Switzerland. She has ongoing communication with Dr. Hornemann. This spring she has conducted her own personal research on how quickly her body adjusts to changing dosages of L-serine in order to determine the lowest effectual dosage. Her goal is to help other patients be able to be more consistent in taking the L-serine and in reducing costs as most patients are having to pay out of pocket for their L-serine. Dr. Hornemann is doing the analysis of her plasma.

Ginger's primary symptom back when she was 11 years old was pain in her right foot leading to casting on numerous occasions throughout her life and a diagnosis of tarsal tunnel syndrome. In her 30's, she started having pain in her hands and had surgery for carpal tunnel syndrome, which was not helpful. Like many others with HSAN1A episodes of severe "lightning" pain has led to a progressive lack of sensation. She has found that taking the L-serine supplement has allowed her to cut back on her pain medication. As her mother died from an accidental overdose, this has been one of the greatest benefits of taking L-serine.

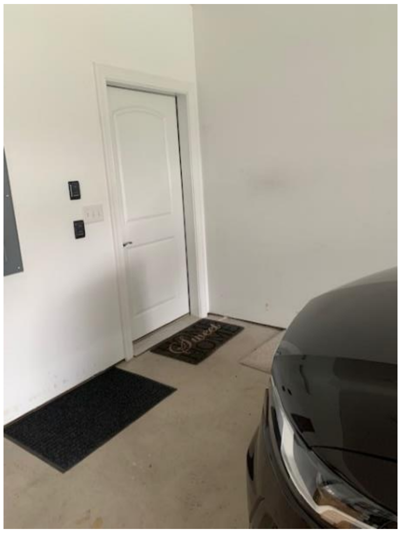
Ginger has three adult children who are asymptomatic and seven grandchildren. As a student at Southwest College of Naturopathic Medicine, she has the opportunity to be an advocate for genetic testing. She lectures in the genetics class on the need for genetic testing in difficult to diagnose cases. Ginger says, "Research needs to be put into clinical practice. Maybe that will be my role someday."

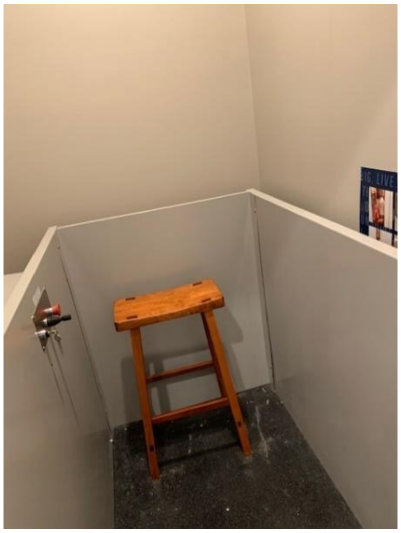
Having watched her mother suffer for so many years with pain that was not managed properly due to improper diagnoses, Ginger has a passion to study, learn, participate in research, and advocate for treatment of HSAN1. She hopes to advance research towards better treatment and perhaps a cure. She says, "My mother's story drives my passion to become a naturopathic doctor, but her story will NOT be my story. I will not let this disease defeat me and I hope to help others find answers sooner rather than 40 years later as it was in my case. No patient should have to suffer for over 40 years without a proper diagnosis. We must push for genetic testing in difficult to solve cases. No patient should have to hear 'no known etiology' ever."

Architectural Adaptations for an Easier Life by David Elston

My wife Jan and I have been very blessed throughout our life together. Recently we were fortunate enough to build a new home. Thanks to the Veterans Administration, a great architect and fantastic contractor we were able to incorporate several adaptations. These changes have helped dramatically in our everyday life :

- Garage floor flush with entrance to home, no steps no ramp
- An open floor plan to incorporate walking
- All light switches lower so you can reach from a chair
- Master bath with large walk-in shower, built in bench seat and hands free shower head and adequate grab bars





- A chair lift (elevator) to get me from the 1st floor to basement
- Wide hallways throughout
- Finished basement with dedicated room for exercise/ PT
- A ramp off the back deck for easy exit to patio
- Exit/entrance door in master bedroom

More things to aid in my independence...

- Custom built closet with open - shelving & accessibility
- Medicine cabinet with open shelves / great for sight and easy access
- Build in microwave at usable height
- Touch faucet
- All cabinets and drawers with pulls that can be opened without a problem.

If anyone would like to visit our home or have any questions, feel free to contact us through the Deater Foundation website at any time.

A GOOD YEAR

by Eric Newcomer, President, Deater Foundation

It has been a good year!

COVID has changed the way we do our jobs, gather with family and friends, go out to eat, and it has messed up the symposium that we were so looking forward to as a way to keep HSAN1 at the forefront of so many doctors and researchers focus from around the world. Then we lost Aunt Louise and Uncle Larue at probably the very worst time of the year to lose a loved one, let alone two of them. While I was thinking about what to write for this year's Deater Foundation newsletter I came across a quote that said: *"It is ironic how often we forget the things worth remembering but remember the things worth forgetting."* How true is that? I know that I'm guilty of it.

It is so easy to get wrapped up in the little things that I have trouble with and forget that I have an amazing wife and two great kids that pick up dropped keys, open the gas cap, button my shirt, and all the other things that used to be so simple. And the dogs are always right there to pick up any food that I drop, but I never seem to get that back.

They made it a good year!

We haven't been able to visit with friends and extended family, but we spent more time together as a family, riding horses, shooting, playing games, and just slowing down. I had more time to watch the birdfeeder and was able to add some new birds to my list like the Brown Thrasher, Rose-Breasted Grosbeak, Indigo Bunting, and a Scarlet Tanger.

That made it a good year!

Considering all that was going on, Cindy and I were able to continue our full-time hours without interruption. I must wear a mask at the college and students are constantly in and out of class quarantining or whatever, but I have been working in the classroom, in-person, and hands-on.

That made it a good year!

Two wonderful people left this earth way too early and that was super hard for a lot of the family, including myself, but I tried my best to think about the good times. Hunting, fishing, and learning to waterski behind the old 15HP Johnson, the nose of the boat pointing toward the sky with Uncle Larue. Listening to Aunt Louise laugh and sing on the big deck at Cedar Haven or the time on Seneca lake one Fall standing in the shallows watching her take off on skis and blue from the cold.

Remembering the things worth remembering made it a good year!

Dealing with HSAN1 in my life every day makes things harder than they need to be for sure, but we have untold numbers of professionals, students, family, and others working now and through the years to find a cure and providing support for non-family members that don't have the history our family does.

That makes it a good year!

In conclusion, I will humbly ask for you to consider donating to the Deater Foundation Inc. so we can continue ***to promote awareness and raise funding to support research on HSAN1, a hereditary sensory neuropathy, with the hopes of finding a potential treatment and cure for this disease.*** Some ways to help include using Amazon Smile (smile.amazon.com) offers the same online shopping experience as amazon.com. But, if you select Deater Foundation, Inc. as your charity of choice, Amazon will donate 0.5% of your eligible AmazonSmile purchases to DFI. Contributions are tax-deductible and can be sent directly to Deater Foundation, Inc. - PO Box 255 - White Deer, PA 17887. Or we do have the ability to accept credit card and direct bank account donations through PayPal and even though they charge 2.9% + a $0.30 transaction fee we feel it is worth the fee structure to provide the convenience of their service.

Thank you for your consideration and have a good year!

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Deater Foundation Inc Treasurer's Report

Balance as of 4/1/20	$12,567.80
Income:	
Contributions 4/1/20 to 12/31/20	16,202.16
Interest 4/1/20 to 12/31/20	1.44
Contributions 1/1/21 to 3/31/21	2,892.90
Interest 1/1/21 to 3/31/21	.72
Total Income	19,097.22
Expense:	
PayPal Service Charges	- 5.35
Total Expense	5.35
Balance as of 3/31/21	$31,659.67

As of 2020 the Deater Foundation has donated $440,000.00 to provide funding for research to find a treatment and a cure for HSAN1.

99.9% of all donations directly support the research, including funding 4 international symposiums gathering experts in various fields to share and expand knowledge of HSAN1.

Dr. Florian Eichler, Massachusetts General Hospital

Dr. Florian Eichler has worked with the Deater Foundation for many years beginning with his time with Dr. Brown in the Day Neuromuscular Laboratory at Massachusetts General Hospital. He was instrumental in the research and implementation of L-serine supplementation for HSAN1. Dr. Eichler has continued to interrogate the pathophysiology of disease related to serine palmitoyltransferase (SPT), examining the impact of dietary changes and diabetes upon the development of neuropathy in mice. Recently he has discovered patients carrying mutations in the small subunit of SPT and is currently studying its impact upon brain development. Besides his academic work, he has together with experts in vector biology co-founded the company SwanBio Therapeutics. SwanBio is a gene therapy company aiming to “rapidly progress the understanding and development of adeno-associated virus (AAV) based gene therapy.” The company focus is on central nervous system disorders, but the basic science also holds hope for peripheral nerve diseases such as HSAN1.

Practical Information on Leg Prosthetics by Christine Deater Christensen

Two years ago, I became a Right Below Knee Amputee (RBKA) due to a bone infection secondary to HSAN1. I have had two types of leg prosthetics in that time. I still wear both types depending on the type of activity I am planning and the weather. The first is called a pin-lock. I roll on a type of sock that sticks onto my leg. There is a long screw-looking pin attached to the end of the sock. That pin locks into an opening inside the socket of the prosthetic leg. This is a great leg if you have a good prosthetist to keep it straight and balanced. It clicks off with the push of a button and then slides back into the lock easily. The two "cons" with this type of leg are 1) you can accidentally hit the lock release and leave your leg in a parking lot, as I have. Then 2) when you do an activity and sweat, there is nowhere for the sweat to go, and my leg constantly slipped out of the sock. The sock would stay locked to the leg but could not stay on my leg. That is both embarrassing and annoying. I slid out of it every day that first summer and I was very frustrated. My prosthetist never offered any solutions, so I found another one. My second prosthetist and I discussed my issues and we decided on a vacuum type prosthetic leg. This type is a bit more involved and heavier but eliminates the worry of me sliding out of the leg. It also feels more natural, like a real leg and not something stuck onto your knee. This type uses a cushion type sock called a liner, to protect the residual limb. Inside this leg there is a pump with a tube. When I pull a sleeve over the socket and inside liner and onto my skin, every step pushes air and sweat out of the tube, creating a tight vacuum seal between my leg and the prosthetic leg. No slippage possible unless I get a hole in the sleeve. When that happens there is no more suction and I replace the sleeve.

Both options are pretty amazing compared to what my dad and uncles had. I can do everything I could pre-amputation.

Another helpful tip from Chris

This mug is for anyone with diminished temperature sensation in their hands. I recently purchased a plastic, double-walled insulated mug at Bed Bath & Beyond. You can microwave it until the liquid inside boils and the cup and handle remain cool. They sell many sizes, in both tumblers and mugs. No more blistered knuckles. I highly recommend this product and you can always pull up a 20% off coupon on their website. Tervis, microwave and dishwasher safe, $10

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In Memory of Louise Adams Hess 1955-2020

By: Carol Ann (Adams) Dorward

Louise was a pioneer in the Deater Foundation's endeavor to find a cause and cure of HSAN1. She participated in various HSAN1 research trials and testing starting, as a young woman, with research at the National Institutes of Health through the most recent L-serine trial. It was Louise's hospitalization with complications of HSAN1 that inspired the establishment of Deater Foundation, Inc. (DFI) in 1990. Louise was regularly present at annual DFI meetings, offering input, perspective, and advice. Her husband Larue provided a constant support to his wife in any way that he could. We are thankful for their commitment to travel and to participate in various aspects of research.

The fourth child born to Lloyd Henry and Verna Ruth Adams, Louise was the baby of the family. She was a wonderful little child who brought much happiness to our home. Even as she grew into adolescence, everyone thought she was adorable.

There is one particular event, I remember so very clearly. Following church one evening, Dad drove us home and went to get Louise out of the car. Why? Because something happened to her in church. You see, she was still "in the Spirit." A young, five years old at the time! Our neighbor Sarah came to help and wondered what was wrong with her. (Imagine Mom trying to explain Louise's condition to Sarah!)

Joy seemed to be Louise's life theme. I have frequently heard others speak about Louise's contagious laughter. That joyful characteristic seemed to seep into everything she did. She was glad to play the harmonica with friends from church. She added spunk to Bible story lessons with children and added that spirited dynamic as she taught them parts to Christmas pieces or plays. Louise was a fantastic storyteller and used her voice inflection to really drive the story home. (Little Orphan Annie was one of my favorite stories.) Louise and Larue both found joy in escaping Pennsylvania winters by snow-birding in Florida and going on cruises with friends and family. Most of all, Louise's joy revolved around her family, especially her grandchildren (whom she adored). Today, I am comforted to know that Jesus, was the reason for, and at the center of, her joy. I believe her involvement in research and her example of living with this disease will bring relief, comfort, and joy to future generations.

Mark your calendars!

The upcoming 32nd Annual DFI Business Meeting is scheduled to be held remotely via Zoom on Friday July 23, 2021 at 7pm (EDT).

All are welcome to attend! If interested, please email us at deaterfoundation@yahoo.com prior to that date for the link information

Deater Family Reunion
July 17, 2021 at noon at Stull, Pennsylvania
Same place as 2018 and 2019 behind Leroy and Isabelle's house at 143 Stonetown Rd, Noxen, PA

- Different Menu: Pot Luck -- everyone bring food for their family plus some extra.
- It all gets put out on the food table for everyone to share.
- Bring your own plates, plasticware, drinks, cups, etc
- People coming from out of state -- buy a bag of chips or cookies etc. to bring and share everyone's food and plates.

Charles Wilson, the Family President, will be putting information on the Deater Family Facebook page.

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