

# Deater Foundation, Inc.

## Newsletter

### HSAN1 Conference 2010

*Submitted by: Ellen Deater Burns, Medical Liaison*

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The Deater Foundation contributed \$10,000 to a newly established Deater Foundation Fund at the University of Massachusetts, Worcester Campus, where the Medical School is located. This money was used to fund the meeting on Hereditary Sensory and Autonomic Neuropathy—Type 1 (HSAN1) held in February in Boston.

The evening prior to the meeting the participants gathered at the Sonesta Hotel for dinner and conversation. Dr. Florian Eichler had recently returned from the Gordon Research Conference on Sphingolipids held this year in Ventura, California. He reported that during the conference there were at least 3 presentations on HSAN1 and at least 4 poster presentations. Dr. Eichler had the opportunity to meet Dr. Walter Holleran, a pharmacist from the VA Hospital in San Francisco, who is known to the Deater family from skin sample testing he conducted on many family members, along with Dr. Fluhr, a dermatologist from Germany, several years ago. Dr. Eichler said Dr. Holleran continues to be interested in the Deater family, and he will contact Dr. Holleran again.

Larry Deater and Rory Robb, Eric and Cindy Newcomer, and Ellen Deater Burns attended the dinner, where the conversation was lively with discussions of cellular biology and physical symptoms of the disease. The interaction continued in hallways and the lounge past midnight as researchers who communicate mainly by e-mail took the opportunity to share ideas and

explore new perspectives on HSAN1. This interaction may have been some of the most valuable of the conference!

The next morning the group was transported to the familiar surroundings of Massachusetts General Hospital's research facility in Charlestown, Massachusetts where Dr. Eichler has his laboratory. The agenda for the day was ambitious and presentations soon exceeded the allotted time with questions and dialog.

#### **Presenting were:**

**Robert H. Brown, Jr., DPhil, MD**, who has worked with the family for more than 20 years, introduced the history of the Deater Family and the identification of the gene for HSAN1. Dr. Brown is the Chair of Neurology and a professor at the University of Massachusetts Medical School. He reminded us that the main signature of the disease is a **small fiber neuropathy**. The genetic basis for HSAN1 is a mutation in the gene that encodes a part of the enzyme serine palmitoyltransferase (SPT). The connection between the enzyme and the destruction of nerve fibers is the subject of research from a variety of viewpoints represented at the conference where basic scientists, clinicians, and family members sat side by side.

**Florian Eichler, MD**, at Massachusetts General Hospital, who attended the Deater Family Reunion in 2009, reported on his work with the

*(More on Page 2)*

#### Reunion

#### Reminder!

Hope to see you

at the 68th

Deater Reunion

on July 17, 2010

at

Butler's property.

## HSAN1 Conference 2010

*Submitted by: Ellen Deater Burns, Medical Liaison*

*—(Continued from Page 1)*

mouse model of HSAN1 and the human clinical trial of serine supplementation. His conclusion is that the disease involves a dysfunctional utilization of the amino acid serine. When the mice are fed a diet high in aniline, another amino acid, they develop the symptoms of HSAN1 at a young age. When the mice are fed a diet high in serine, the symptoms are minimized and in some cases have disappeared. Neurons, or nerve cells, do not produce serine on their own. Serine must be available and transported into the body of the nerve cells. During the clinical trial family members with HSAN1 took supplemental serine in doses of either 200mg/Kg or 400 mg/Kg. The levels of certain types of lipids called deoxysphingoid bases (DSB) that are known to be markers of HSAN1 decreased significantly with high dose supplementation. After the conclusion of the study, the levels of the DSBs returned to the pre testing levels.

The question was asked, "What is wrong with SPT in HSAN1?" and the next researchers proposed theories to answer that query.

**Thorsten Hornemann, PhD** from the University of Zurich in Switzerland, provided an update on the deoxysphingoid bases in HSAN1. He has been working closely with Dr. Eichler in analyzing the results of the clinical trial. He noted that deoxysphingolipid levels are elevated in plasma and peripheral nerve tissue, that they are the plasma markers for HSAN1, and that in the laboratory in culture medium, deoxysphingolipids are toxic to nerves.

**Hyujung Park, PhD**, of Boston University spoke on SPT kinetics. This is the study of how different conditions can influence the speed of chemical processes. This information can lead to a better understanding of the

mechanism of the chemical reaction. The SPT enzyme reacts differently to different circumstances.

**Annelies Rotthier, PhD**, at the University of Antwerp, Belgium, presented a broader look at inherited peripheral neuropathies, including Charcot-Marie-Tooth and the variants of Hereditary Sensory Neuropathy. She introduced a new mutation suspected to be HSAN1. The other meeting participants, however, discounted this single case as unlikely to be a true HSAN1.

**Teresa Dunn, PhD**, Uniformed Services University of the Health Science, Washington DC, has been working with Dr. Brown and Dr. Eichler for several years. She works primarily with yeast and looks at functions within the cell. Her work investigates the SPT processes at a cellular level.

**Jonathan Lowther, PhD** works with Dominic Campopiano at the School of Chemistry, University of Edinburgh, Scotland. They combine chemistry, microbiology and immunology to study proteins and enzymes and ascertain the structural characteristics and mechanisms of encoded proteins. By visualizing the structural model, other scientists can better understand how the protein can interact with other elements within the cell.

**Peter Novak, MD, PhD** at the University of Massachusetts Medical School explained the process of autonomic testing in neuropathies. Tests include several small punch skin biopsies to visualize sensory and autonomic peripheral nerves in the skin, a test of sweat production, a tilt table test, and measurement of the heart rate with various tests. Dr. Novak showed actual test results from the first participant in what is planned to be a



**Ellen Deater Burns**  
*Medical Liaison*

*DFI File Photo*

*"This family is  
strongest standing  
together; and, with  
faith, we can move  
mountains!"  
—Ellen D. Burns*



**DFI**

*Continued on Page 3*

## Pictures from Last Year's 67th Deater Reunion

*July 18, 2009 Submitted by: Carol Dorward*

Do you remember last year's reunion? It was a beautiful day with the temperature between 70 and 73 degrees all day. About 176 people attended, some from as far away as Northern California, Oregon, Tennessee, and Colorado.

Special guests included Dr. Eichler and his assistant, Ourania Giannikopoulos, who were there, getting acquainted with the family—just prior to the initiation of the short term clinical study which was scheduled for the following day.



On July 17, 2010, we will, once again, gather together at Butler's property for the 68th Deater Family Reunion. We look forward to the fellow-

ship with family we seldom get to see, as well as the good food, singing, games, etc.

Mark your calendar! Hope to see you there.



*Mark your calendar  
for this year's  
reunion:*

*July 17, 2010*

*DFI File Photos*



## HSAN1 Conference 2010

*Submitted by: Ellen Deater Burns, Medical Liaison—(Continued from Page 2)*

new study of HSAN1.

**Ann Louise Oaklander, MD, PhD** at Massachusetts General Hospital said the Deater family changed her perception of HSAN1. She explained that small nerve fibers do not just sense pain but prevent and respond to injury. The nerves involved in HSAN1 can most easily be measured by viewing a skin punch biopsy, as the epidermis of the skin contains the small nerve fibers. From a clinician's viewpoint, Dr. Oaklander talked about neuropathic pain which she says is caused by the remaining neurons firing inappropriately, and about the loss of enervation leading to loss in bone regulation, thinning of the skin and blister formation, and deactivation of sweat glands.

**Nazem Atassi, MD** at Massachusetts General Hospital spoke about a consortia and trial for rare diseases. This model is in place for the study of Amyotrophic Lateral Sclerosis (Lou Gehrig's disease) with 92 sites of expertly trained clinicians. In this situation Massachusetts General Hospital serves as the coordinator and the State University of New York is responsible for monitoring and outcomes.

**Garth Nicholson, PhD** from the Concord Clinical School, The University of Sydney, Australia spoke briefly to assert that HSAN1 should be classified as a

*Garth Nicholson, PhD  
from the Concord Clinical  
School, The University of  
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fied as a Charcot-Marie-  
Tooth disease.*



**DFI**

*Conclusion on Page 6*



# Martha Francis Deater Herring Dennis —That is our Mother

*Submitted by: Bobbie Herring Kirk*

Martha, better known as Mickey, and not to be confused with the famous mouse, was born on September 15, 1931, and is still young at heart. She is next to the youngest of twelve. She grew up in Noxen, PA.



\*1

Mickey married our father in November 1951. Bobbie arrived in 1952; Cindy was born in 1955; Tim arrived in 1958. Mickey lost a baby, Mary, in 1960. Then our blond ambition and baby of our family, Lisa, arrived in 1965.

In 1955 Mickey moved to Johnson City, NY. She worked for Endicott Johnson making shoes and sneakers. We also got to live next door to Aunt Laura Mae and Uncle Russell on the second floor.

In 1961 we moved about 30 miles from Johnson City to Candor, NY. Mickey lived there until 1975. Shortly after Lisa arrived, Mickey was left to face some challenges when our father left for the "FAR COUNTRY." She worked at NCR (National Cash Register) in Ithaca, NY. There she did piece work. She seemed to enjoy her job and also worked in the kitchen at a restaurant/bar in Owego, NY some evenings. She kept pretty busy with 2 jobs, a house to maintain and 3 kids to oversee. Yeah, we pretty much were on our own a lot. However, Tim and I turned out all right. We're still wondering about Lisa (tee hee!)

Lisa remembers mom not being afraid of anything or anyone for that matter. She remembers as a kid opening up the back door in NY to let the dog or cat in and in comes a possum (not once but several times). Those things are mean and ugly! Mom would just grab a bat and beat them to death just like that.

The good times always outweighed the bad times. I remember her sitting out on the porch in Candor singing and playing her accordion. We remember riding in the back of her VW convertible with my cousins Nancy and Tonja. What a hoot!



\*2

In 1975 Mom married Donald Dennis and moved to Berwick, PA, where she now resides. While there she had her own cleaning business for some time. For several years she and Don had a camper and would travel around to parks camping out.

Lisa always thought mom wasn't afraid of anything until one day she was in the garden with our little stepbrother, Scotty. Lisa had just taken another load of clothes out to hang. Pretty soon she heard Scotty screaming and running towards the house and mom is just screaming and running in place as fast as she could go (yet she wasn't moving an inch) all because a snake crawled across her foot. Well let me tell you, I on the other hand am afraid of EVERYTHING but I knew I had to go get her. I've never seen her run so fast. (Ha ha).

Mom has had numerous health issues these last several years and keeps both my brother and sister on their toes. I hope they know how much I appreciate what they do for her. Several years ago, over a period of time she was given numerous pints of blood to the point that she jokingly said she didn't know what color she was anymore.

I also heard that when the town decided to put a sewage plant on her property and proceeded before she had heard back from her lawyer that she went out and confronted the workers and told them they needed to stop working. Well, the workers did not stop; so she said, "Listen, I have a lawyer and a gun." – and they made tracks fast then. They also called the cops on her. Apparently, the cop said, "Martha, you can't be threatening people"—to which she said, "I didn't threaten anyone—I only said 'I have a lawyer and a gun' which is the truth! I didn't say I was going to shoot anyone."

*This issue of  
the Deater Foundation  
Newsletter  
is  
in Honor of  
Martha Francis  
Deater Herring  
Dennis*



*Martha Francis Deater  
Herring Dennis*

*Photo courtesy of  
Bobbie Herring Kirk*

**Born: September 15, 1931**

**Married: Robert LaRue Herring  
(1951)**

**Children:**

**Bobbie Herring Kirk**

**Cindy Sue Montross Gitschlag**

**Tim LaRue Herring**

**Mary Herring**

**Lisa Anne Herring Hunsinger**

**Married: Donald Dennis (1975)**



**DFI**

*Continued on Page 7*



## DFI Receives Grant from Enterprise Holdings Foundation

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*Submitted by: Carol Dorward and Ellen Burns*

The Deater Foundation, Inc. (DFI) headquartered in northeast Pennsylvania, was recently awarded a \$2,500 grant from Enterprise Holdings Foundation. Deater Foundation, Inc. is a non-profit organization that funds medical research for HSAN1, a hereditary, debilitating, disease that affects the nerves and muscles. HSAN1 has affected families in the United States and other countries for generations.

Jon Ellsworth, who has worked for Enterprise Rent-A-Car for the past thirteen years, submitted the application in early Spring and was notified of the award on May 25, 2010. Jon

says that he is proud to work for a company that gives back to its employees through generous philanthropic gestures for causes such as the medical research that the Deater Foundation supports.

Deater Foundation, Inc. has helped to make HSAN1 research possible for several decades now, and, combined with other funding sources (such as this generous grant from Enterprise Holdings Foundation), is making great strides toward a treatment for HSAN1. For more information about HSAN1 and/or the DFI organization, go to [deaterfoundation.org](http://deaterfoundation.org).



**Jon Ellsworth**

*Photo used with permission*

*Editor's Note: Deater Foundation, Inc. is grateful to Jon Ellsworth for continuing to pursue this grant year after year. This is the fourth year that he has submitted an application and the fourth year that DFI was awarded a generous grant. Thank you, Jon!*

*Deater Foundation, Inc. is equally grateful to Enterprise Rent-A-Car and the Enterprise Holdings Foundation for their support of DFI these past four years. Deater Foundation is honored to know that Enterprise Rent-A-Car and the Enterprise Holdings Foundation have considered DFI to be worthy of their support and generosity. Deater Foundation is committed to being good stewards of all gifts in order to expedite the medical research for a treatment/cure for HSAN1.*

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## Initiation of a New Study

*Submitted by: Ellen Deater Burns, Medical Liaison*

In January this year the Deater Foundation submitted a letter in support of a funding request for a new study: "Accumulation of Atypical Sphingolipids in HSAN1: A Therapeutic Target?" In February, the Deater Foundation supported an international symposium to discuss the progress made to date and the direction for future studies in the search for a treatment or cure for HSAN1. Two questions emerged:

- \* Will the serine treatment that was trialed last summer have a lasting effect on the symptoms of the disease?
- \* What specific outcomes need to be measured to determine if the treatment is effective?

The family had, over the years, collected information about such things as the age when an individual first dis-

covered the disease, the sex and generation in the family, and, recently, dietary and life-style habits and blood tests. There had, however, not been a coordinated objective study of the progression of the disease. In preparation for the proposed new study, several physiologic tests were performed on two affected family members. These include autonomic testing, skin biopsies to study peripheral nerves, nerve conduction studies and a clinical examination.

Application for funding for these studies has been applied for but not yet awarded. In spite of this, Dr. Bob



*Continued on Page 8*

## HSAN1 Conference 2010

*Submitted By: Ellen Deater Burns, Medical Liaison—Continued from Page 3*

Charcot-Marie-Tooth disease. HSAN1 clearly has elements of both sensory and motor disorder, and all nerves have autonomic fibers, so the “autonomic” part of the current designation is misleading.

### **Final Comments:**

In addition to the presenters, there were numerous other participants. Alex McCampbell, who developed the HSAN1 mice model and now works for Merck was an active contributor, as were PhD students and people in post doctoral programs bringing fresh perspectives to the conversation.

In the discussion following the presentations the essential question was, “What’s next.” Dr. Robert H. Brown, Jr. pointed to the initiation of a longitudinal study (mentioned above in Dr. Novak’s presentation). This study

could start small with the autonomic battery of tests, neurological examination, and background information (a “natural history” of the disease). Various time frames were proposed, from 12 weeks to one year or longer. This could be the core for a larger study that could be conducted at various sites using the same protocol for the families already being studied in England, Australia, and the United States. Dr. Eichler is the most likely person to write the protocol for the study. Dr. Brown talked of presenting results as soon as possible at a neurology conference and possible sources of grant funding were discussed.

Larry Deater and Eric Newcomer urged the scientists and clinicians to act quickly. Eric reminded them that, “Every day that you delay, I get worse.” We all left with a renewed sense of commitment and hope that new discoveries are on the horizon.

*We are hopeful  
that  
“new discoveries  
are on  
the horizon!”  
—Ellen Deater Burns*

## DFI Treasurer’s Report (June 1, 2009, to May 31, 2010)

*Submitted by: Nancy Newcomer, Treasurer*

### DFI Statement of Account

Balance as of 6/1/09	\$35,498.83
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#### Income:

Contributions 6/1/09 to 12/31/09	11,868.59
Interest 6/1/09 to 12/31/09	17.04
Contributions 1/1/10 to 5/31/10	2,830.00
Interest 1/1/10 to 5/31/10	3.33

#### Expenses:

Massachusetts General Donation (July 2009)	- 25,000.00
U Mass Donation (January 2010—for Feb. Conference)	- 10,000.00
PayPal Service Charges	- <u>5.30</u>

Balance as of 5/31/10	\$15,212.49
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Nancy J. Newcomer  
Treasurer



DFI



# Martha Francis Deater Herring Dennis—That is our Mother

*Submitted by: Bobbie Herring Kirk—(Continued from Page 4)*

My mom loves game shows and putting puzzles together. She likes to gossip and sing; and she is an EXCELLENT “cinnamon bun” baker. She is also very OUTSPOKEN—No matter what or who, she tells it like she views it!

Mom, I love you MORE!—Bobbie

I love Mom BEST!—Lisa *(no matter what my stupid sister says!)*

*With love  
from all of  
your children  
and grand-  
children.*



Tim, Bobbie, Lisa and Mickey  
—Photo courtesy of Bobbie Kirk

Web Credits for Clip Art on Page 4:

\*1—<http://www.lincah.com/wp-content/uploads/2009/12/2010-Volkswagen-New-Beetle-Final-Edition-Convertible-Front-Angle-View-588x379.jpg>

\*2—<http://blog.beliefnet.com/moviemom/mickey-mouse-10.jpg>

*Keep on keeping on!*

## Gift Certificate Reminder

—Submitted by Rory Robb Deater

For Your Consideration:

A gift to Deater Foundation, Inc. may be the perfect gift for someone, to honor someone, to remember someone.

Give a donation:

- \* To celebrate a birthday
- \* In honor of Mother's Day or Father's Day, for the parent with HSN-1 (or without)
- \* In appreciation
- \* For Christmas, for the hard-to buy-for person
- \* In memoriam

You may request a certificate, suitable for gift giving, to present to the recipient. To do so, contact Nancy Newcomer, Treasurer, P. O. Box 255, White Deer, PA 17887 or email her at: [njnewcomer@yahoo.com](mailto:njnewcomer@yahoo.com)

## “Grandma Mickey”

*Submitted by: Kristen Kirk-Paladino*

Our Grandma Mickey is truly one of a kind. Our grandma's heart is bigger than the universe. Her laughter fills up a room. She has opened her home to many from her church in time of need. That sometimes meant we got stuck with the couch when we would come and visit. Growing up, we have seen her spirit and charity to others, and she has handed that down to all of us. She taught us that the key to a better world is to “Pay it Forward.” Then there is the “truly one of a kind” that she is: Like spending time with her in the summers and finding a rabbit for dinner in the sink—*(Thankfully she had hot dogs for us)*—To telling us it was time to shave her whiskers. I hope she doesn't pass that down to us, but I think she already has.

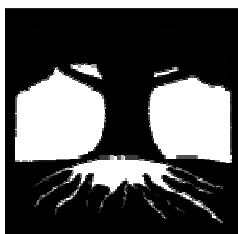
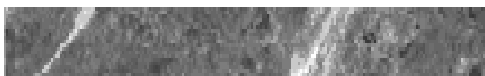


Grandma Mickey  
enjoying a pickle on a stick at  
Knoebel's

—Photo courtesy of  
Kristen Kirk-Paladino



DFI



**DFI**

**Deater Foundation, Inc.**

**P. O. Box 255**

**White Deer, PA 17887**

*Remember that your  
contributions to DFI are  
tax deductible.*

*Mail contributions to:  
Deater Foundation, Inc.  
c/o Mrs. Nancy Newcomer  
P. O. Box 255  
White Deer, PA 17887*

**We are on the Web!**  
[deaterfoundation.org](http://deaterfoundation.org)

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**Join our Facebook Group:  
Search for Deater Foundation**

## **Initiation of a New Study**—(Continued from Page 5)

***Submitted by: Ellen Deater Burns, Medical Liaison***

Brown, who has championed the study of the Deater family for many years, and Dr. Florian Eichler, a more recent supporter, have decided to go ahead with the study now. Four additional affected family members have volunteered to participate in June. We anticipate that, with appropriate grant funding, every affected member of the family who is interested will be tested.

The doctors would like to increase the size of the group being tested by including other people with HSAN1 in England and Australia. They have been in touch with other researchers, notably Drs. Mary Reilly and Garth Nicholson, who have both been studying HSAN1 families for many

years.

This July, at the time of the Deater Family Reunion, Drs. Brown and Eichler will be presenting Amino acid substrate selectivity alters desoxysphingoid bases and disease severity in HSAN1 at the 12<sup>th</sup> International Congress on Neuromuscular Diseases in Naples, Italy. This is a report on the serine study and related research. We are fortunate to have such dedicated researchers working on a treatment and cure. Dr. Mary Reilly's presentation at the same International Congress asks if a treatment for HSAN1 is near. We pray that with the support of families and researchers around the world, the answer is "YES!"

**Not on our Mailing List? New Address?**

Notify: Carol Dorward

at [carol\\_dorward34@yahoo.com](mailto:carol_dorward34@yahoo.com)

or [info@deaterfoundation.org](mailto:info@deaterfoundation.org)

*of your current home address and/or email address*



**DFI**