

Deater Foundation, Inc.

Newsletter

Research Progress Report 2012

Submitted by: Ellen Deater Burns, Medical Liaison

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Shortly after the Deater Family Reunion in July, 2011, Larry Deater and Ellen Deater Burns traveled to Boston to meet with Dr. Eichler. We shared some concerns that the Deater Foundation had contributed \$30,000 in January to the proposed research on Hereditary Sensory and Autonomic Neuropathy Type 1 (HSAN1) and it seemed to us that the movement toward a study involving serine supplementation effects on people with HSAN1 was painfully slow.

Dr. Eichler had constructed a placebo controlled double blind study to meet the funding qualifications of large organizations such as the National Institutes of Health (NIH) and the Food and Drug Administration (FDA). Unfortunately, funding from those sources, which looked promising initially, has not materialized. The smaller trial of 5 persons affected with HSAN1 receiving serine supplementation was also off to a slow start. One of the criticisms that Dr. Eichler received from the FDA was the high cost of the L-serine. His team "shopped around" for a lower priced, high quality product. The L-serine had to be vetted by the pharmacists at Massachusetts General Hospital (MGH) to meet the same quality standards as the L-serine used in the first 10 week trial. Another hurdle to overcome was finding a suitable laboratory for the follow-up blood draws. In the end, the participants agreed to have their blood drawn and

shipped to MGH via FedEx for analysis. There were continuing challenges with the MGH Institutional Review Board, which must review and approve all human research trials at MGH. Thanks in large part to the efforts of Jessica Pan, Clinical Research Coordinator in Dr. Eichler's laboratory, the study was approved.



Ellen Deater Burns
Medical Liaison

DFI File Photo

In December, 2011 an article was published in the Journal of Clinical Investigations: "Oral L-Serine Supplementation in Mice and Humans with Hereditary Sensory Autonomic Neuropathy Type 1." The authors, in addition to Dr. Eichler, are: K. Garofalo, A. Penno, BP Schmidt, HJ Lee, MP Frosch, A. von Eckardstein, RH Brown, and T. Horne-mann. The article reports on the mice study and the L-serine supplementation trial involving 14 Deater family members.

In the same publication, Steven S. Scherer, M.D., Ph.D., professor of neurology and researcher at the University of Pennsylvania, wrote an article titled, "The debut of a rational treatment for an inherited neuropathy?" He presents a succinct overview of the research to date in HSAN1 and concludes that "unraveling the cause of neuropathy in individuals with HSAN1 will require more study."

(Continued on Page 2)

DFI Receives Grant from Enterprise Holdings Foundation

—Submitted by: Jon Ellsworth

Another year has passed and here we are beginning the Summer of 2012. I can tell you it's going to be a HOT one here in SW Florida. As I write this at the end of May, we are already having 95 degree days.

This year I was again able to apply for a grant through the Enterprise Holdings Foundation, on behalf of DFI. We recently found out that DFI has been awarded a \$3500 grant to continue to help fund research towards treatments and an eventual cure for HSAN1. Enterprise Holdings is the parent company for Enterprise Rent-A-Car, Alamo Rent A Car and National Car Rental. I've had the privilege to work for Enterprise for the last 15 years. This is the 6th year that Enterprise has graciously approved a grant donation for DFI. As in years past, I encourage everyone to check with your employer's Human Resource department and see what types of opportunities exist that could benefit DFI: grants,

donations, payroll matching – to name a few.

Research for a cure is especially important to me and my family. As most know, my mom [Eileen Ann (Deater) Ellsworth] has been dealing with HSAN1 since she was about 18 years old. I, too, have been working through symptoms for many years; and now my oldest daughter has had some minimal numbing in her feet. We all face this head on, and we know that we have a God who will one day give us perfect healing. But in the meantime, we so much support and appreciate the work that doctors and technicians are doing and are grateful that God has given them the skills to do it!

Thanks for your continued support of DFI!!



*Jon Ellsworth and Family
—Hope, Alexis, Japeth,
Abigail and Josiah—
[Photo used with Permission]*

Editor's Note: Deater Foundation, Inc. is grateful to Jon Ellsworth for continuing to pursue this grant year after year. We applaud you for your efforts. Jon, we appreciate you and thank you so much.

Deater Foundation, Inc. is equally grateful to Enterprise Holdings Foundation for their faithful support of DFI. Deater Foundation is honored that Enterprise Holdings Foundation has considered DFI to be worthy of their support and generosity year after year.

Research Progress Report *(Continued from Page 1)*

—Submitted by: Ellen D. Deater Burns, Medical Liaison

In January, 2012, Dr. Eichler was informed that his large study was not likely to be funded in the spring. He plans to revise his application to make the proposal stronger and re-submit to the FDA in October of this year.

In a February analysis of the blood samples drawn some months previously, it was noted that the potassium levels were alarmingly high. Supplementation was stopped until it was determined that the elevated levels were due to an aberration

of the shipping and storage of the blood samples, as they became overheated in the process of moving the samples about. Electrolytes had not been measured in the initial 10 week trial. The study continues with the participants in the study being followed quarterly through blood testing and analysis.

The studies with the mice also continue. "One important insight that is worth reporting," Dr. Eichler says, "is the reaction of our HSAN1 mice to a high fat diet. Unlike wild

type mice, the HSAN1 mice do not put on weight and start to have severe intestinal motility problems. These problems are related, but not exclusively, to pathology of the pancreas that is not seen in wild type mice or on other diets. All this happens very quickly, within a month, and tells us that beyond the amino acids there are other culprits to exacerbate



DFI

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Pictures from Last Year's 69th Deater Reunion

July 16, 2011—Submitted by: Carol Dorward



DFI File Photos



Mark your calendar for this year's reunion: July 21, 2012

Research Progress Report 2012 (Continued from Page 2)

—Submitted by: Ellen D. Deater Burns, Medical Liaison

bation in HSAN1. Now, the high fat diet (60% calories from lipids) is nothing a human would be exposed to but the fact that the HSAN1 mice react to it but not the wild type mice *is* significant. This lends more credence to a dormant autonomic dysfunction in HSAN1. I think the cumulative lipid content of the human diet could play a role in the lifelong disease.”

A young investigator at MGH is writing up the natural history of HSAN1 based on the surveys the family provided. Another family has also filled out the survey. The investigator has taken a special interest in the disease.

The abnormal lipids found by Thorsten Hornemann in HSAN1 are also found in diabetes patients but in lower amounts. The teams

are exploring whether there is a common thread between the two diseases.

Further information, including graphs of the results of the 2009 L-serine supplementation study with Deater family members, can be found at the MGH Eichler Lab website, <http://eichler.mgh.harvard.edu/hsan1/>

Dr. Eichler continues to apply for funds to proceed with clinical trials. However, in the current economy it is hard to predict whether funding will be forthcoming. Deater Foundation funding remains an important support for the study of this disease. The Foundation sent a letter of support in June to Dr. Eichler for a modification to the second year of the proposed double blind randomized study that he has

devised, and for which he continues to seek funding. The new grant proposal is going to the National Institute of Neurological Disorders and Stroke, a branch of the National Institutes of Health. If funded, the study would not start until the spring.

Dr. Eichler plans to resubmit the grant to the FDA this coming fall. He got an excellent score on the first try and funding seemed imminent but did not materialize, as resubmissions were considered in the same pool as new applications. We appreciate Dr. Eichler's tenacity and persistence to get financial support for this important study. We pray that the current trial and next study will lead to a viable treatment of HSAN1.



DFI

DFI Treasurer's Report (June 1, 2011 to May 31, 2012)

Submitted by: Nancy Newcomer, Treasurer

DFI Statement of Account

Balance as of 6/1/11	\$ 8,009.51
<u>Income:</u>	
Contributions 6/1/11 to 12/31/11	15,911.85
Interest 6/1/11 to 12/31/11	7.48
Contributions 1/1/12 to 5/31/12	13,531.47
Interest 1/1/12 to 5/31/12	2.92
<u>Expenses:</u>	
Network for Good Service Charges	- 50.81
PayPal Service Charges	- <u>1.40</u>
Balance as of 5/31/12	\$37,411.02



Nancy J. Newcomer
Treasurer

Foundation Support

Submitted by: Ellen Deater Burns, Medical Liaison

The Deater Foundation was established to provide funding for research into Hereditary Sensory and Autonomic Neuropathy Type 1 (HSAN1), the inherited disease that has now afflicted at least 5 generations of the Deater Family. Sixth generation children are at risk. Monies from the support of the Foundation have, over the years, partially paid the salaries of laboratory technicians, been instrumental in the discovery of the genetic mutation that causes the disease, assisted in the development of the HSAN1 mice, and funded international research conferences that encouraged lasting collaborations among researchers.

The largest single grant ever given by the Foundation was \$30,000. This most recent grant, which almost exhausted the resources of the Foundation at the time, was given last year to support research into the effects of providing a dietary supplement, L-serine, to people with the disease. Often, Deater Foundation grants have provided "seed money" to encourage larger grantors to also give to support

the ongoing research. This grant is specifically paying for L-serine supplementation and the processing of blood samples for people in a small clinical trial. It is hoped that by providing supplementation with serine, the body will be more able to utilize that essential element, and consequently the disease may cease to progress. Potentially, supplementation may even reverse the symptoms of the disease.

These funds have now been exhausted. The study will continue for several more months, with quarterly analysis of blood samples. There will also be extensive neurological testing at the completion of the study to compare nerve function after a year of supplementation with the studies done prior to the trial.

Your donation NOW could be critical to the development of a treatment for HSAN1! The study participants report that they feel better while taking the supplement. We may be on the



brink of a significant turning point in this disease! Please consider giving to the Deater Foundation. Give what you can, and then give a little more. ***Your contribution may mean the difference in a few years between walking and a wheelchair, for a child today.***



DFI

Recycling of Old Cell Phones and Printer Cartridges to Benefit DFI

—Submitted by: *Tami Newcomer Murphy*

Help Deater Foundation, Inc. by continuing to donate your used cell phones and empty laser and inkjet printer cartridges!

Recycling these unwanted items can help the environment as well as raise money for DFI. You can also check with your local businesses, place of employment, school, church, etc. about donating their unwanted items as well. All brands and models accepted.

For more information, contact Tami Murphy: (610) 873-1310 or newcomert@hotmail.com ... and **plan on bringing your items to the reunion in July!** *Gary and Nancy Newcomer will be glad to accept these items from you at any time throughout the year and at the reunion, of course.*

Thank you for your support this past year. Let's continue to take advantage of this opportunity to support Deater Foundation, Inc.



*Tami N. Murphy, Vice President
and her daughter, Averi*



Gift Certificate Reminder

—Submitted by *Rory Robb Deater*



A Couple More Reunion Photos

For Your Consideration:

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

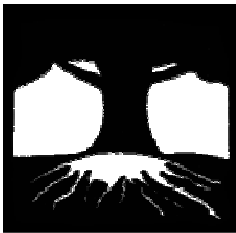
You may wish to give a donation:

- * To celebrate a birthday
- * In honor of Mother's Day or Father's Day, for the parent with HSN1 (or without)
- * In appreciation
- * For Christmas, for the hard-to buy-for-person
- * In memoriam

To request a certificate suitable for gift giving to present to the recipient, contact **Nancy Newcomer, Treasurer, P. O. Box 255, White Deer, PA 17887** or email her at: njnewcomer@yahoo.com



DFI



DFI

**Deater Foundation, Inc.
P. O. Box 255
White Deer, PA 17887**

Email: deaterfoundation@yahoo.com

*Your contributions
to DFI are
tax deductible.*

*Mail contributions to:
Deater Foundation, Inc.
P. O. Box 255
White Deer, PA 17887*

We are on the Web!
deaterfoundation.org

*Join our Facebook Group and Cause
by searching for Deater Foundation*

Appreciation for Financial Donations

Submitted by: Carol Dorward, President/Secretary

Deater Foundation, Inc. is grateful to Enterprise Holdings Foundation for their faithful support by awarding yearly grants to Deater Foundation over the past six years. We are blessed to have had the Enterprise Holdings Foundation consider DFI to be worthy of their faithful support and generosity. It may surprise you to know that we have received a total of \$16,000 in grants from EHF since 2007. That's tremendous! And we so appreciate Jon Ellsworth for making the application each year for these grants.

We've been blessed with other special donations as well. Thanks to Brad Fleeger, DFI received a generous donation this year from Melbourne Lodge No. 143, Free and Accepted Masons of West Melbourne, FL. That was a pleasant surprise. Thanks, Brad, for your involvement in making this happen.

Early in 2012, another unusual, generous donation came in the form of a bequest from the estate of Ruby Scouten.

Perhaps the knowledge of these special donations will inspire others to consider the possibility of supporting DFI financially via their employers' programs, benevolent organizations that they belong to, or by making DFI a beneficiary in their estate planning.

Whether you give to DFI creatively via estate planning, employer programs, United Way, etc. or via yard sale proceeds, faithful monthly or yearly donations, etc., we appreciate each donation.



Carol Dorward
President/Secretary

Deater Foundation, Inc. is committed to being good stewards of all gifts in order to expedite the medical research for a treatment/cure for HSN1.

Not on our Mailing List? New Address?
Notify: Carol Dorward
at carol_dorward34@yahoo.com
or deaterfoundation@yahoo.com
of your current home address and/or email address



DFI