DEATER FOUNDATION, INC.

June 30, 2006

Volume 8, Issue 1

Deater Foundation, Inc.

Disease Research Report

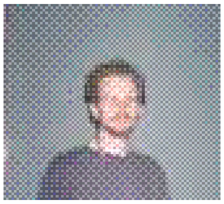
Submitted by: Ellen Burns, Medical Liaison

Inside this issue:

Reunion Reminder!

Hope to see you at the 62nd Deater Reunion on July 15, 2006 at the Grove in Stull.

Paper Presentation: Mouse Model for Hereditary Sensory Neuropathy Type I



Alex McCampbell, working in the Day Laboratory for Neuromuscular Research, presented a paper on hereditary sensory neuropathy in late 2004 at the Society for Neuroscience. Alex has been working for several years with Dr. Bob Brown, Jr., who is the Director of the Day Lab. Alex has been working to develop a strain of mice who demonstrate the defective gene found in the human form of HSN-1. Once referred to as the "Deater Disease," HSN-1 is now known to be the most common hereditary disorder primarily affecting peripheral sensory neurons.

The gene with the mutations associated with HSN-1 is identified as **SPTLC1**. Genes make proteins; proteins make enzymes. **SPTLC1** encodes one subunit of the serine palmitoyltransferase enzyme (SPT). This enzyme is known to affect the production of a fatty substance (glycosyl ceramide) in the body. Alex altered the genetic makeup of mice in several ways. He created transgenic mice. One group of mice have the mutated gene that is associated with HSN-1 added. The other group of mice was altered to over express the normal gene. The two groups were created to try to determine if the nerve damage in HSN-1 is caused by the SPT, which might mean SPT has a toxic, or poisonous effect, or if the damage might be caused by a decrease in the SPT activity, caused by the presence of the mutant protein.

Although mice and people share most of the same genetic structure, they are obviously not the same, and it is always uncertain if things are going to work the same way in mice as they do in people. For one thing, Alex had to find out if the mutant gene was reproduced when the mice reproduced. This was successful, and prepared the way for additional study. Then Alex had to determine if the genes added to the mice were functioning. The mice with the mutant gene did express the gene and did show significantly decreased activity of SPT. Some of

DFI

(Continued on page 3)

Of Interest…"A 1937 Letter from Harvey Deater"

Submitted by: Beulah Womer

Thursday Nite
Nov. 18

Dear Folks:

We came in the hospital yesterday about nine o'clock but it was eleven before we were admitted. We missed out on dinner but were xrayed from our chest and had supper. They feed swell here.

Lights out at nine PM then they get us up at 4 in the morning we wash up then wait until seven for breakfast, dinner at 11 & supper at 5.

Today they took three blood tests, a urine test and this afternoon they photographed our feet seperatley (sic) and together.

The Dr. that is Rugh(?) was in this afternoon and he knows practically everything about the disease but he does not know what causes it or how to cure it.

He said if we stayed off our feet completely it would heal up, but the first time we walked far on them they would be sore again. He does not want to heal them that way so we are allowed to walk around. They gave us pyjammies (sic) and a robe and we have our shoes.

Tomorrow we have another xray head and all a complete body xray. I don't mind it so much now and I don't think Russell does either but up to noon today from noon yesterday it seemed like a year.

There are 24 in the ward, all odd cases and the fellows are all nice. The present bunch of nurses are swell, but some of them get changed tomorrow.

Alice brought us over and stayed until we were located in the ward about 2 o'clock. Then she came in today and brought us candy oranges nuts & grapes. I don't know who is going to eat them as she said she would be back Sat. and bring more.

We are both fine but have no idea how long they will keep us. We have been examined by a hundred or more Drs. already.

Hoping everyone & everything is OK up there will say goodbye.

With Love,

Harvey & Russell

Harvey Deater
Mens Special Ward
Jefferson Hospital
Philadelphia, PA

This letter was found among Grandma's things. According to Aunt Boot, the letter had been mailed with a 3-cent postage stamp in 1937!

Let's see... In 1937, Harvey would have been about 26 years old and Russell would have been about 24 years old.

Website Comments, Suggestions, Questions, etc.

Contact Dixie Dorward:

ddorward@deaterfoundation.org

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

Pictures from Last Year's 61st Deater Reunion

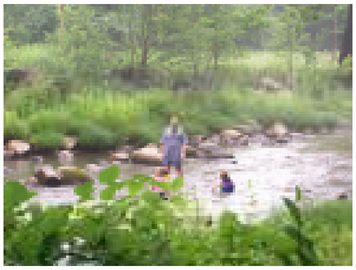
July 16, 2005

Once again, we will meet for good food, fun, and fellowship this summer on **July 15, 2006** at the Grove in Stull.

Hope to see you there. We always have a great time.

Do you recognize the new bridge over Bowman Creek?...

...and one of the favorite swimming/wading spots?



Disease Research Report

(Continued from page 1)

the mice with the mutation were smaller than the normal mice.

The mice with the mutation showed decreased sensitivity to temperature sensation as they aged. The mice did not show any motor problems, or loss of function. However, because of the success of the development of the mice, it is thought that the mice may be a useful model of HSN-1. The researchers learned a lot from the development of the mice. More studies need to be done to take the research into HSN-1 further. The mice will be available for other researchers. Alex McCampbell has concluded his post-doctoral time with the Day Lab and has taken a job with Merck Laboratories; but Dr. Brown has hired a new technician to continue Alex's work, which will also be pursued in the Day Laboratory by Dr. Florian Eichler, a pediatric neuropathologist on staff at the MGH and Harvard Medical School.

Dr. Florian Eichler, a pediatric neuropathologist on staff at the MGH and Harvard Medical School is now pursuing the research in the Day Laboratory.

In Honor of June Deater Sorber

Submitted by: Claude A. Sorber

June Ellen Deater Sorber

Born May 26, 1922

7th Child of Alvin & Ellen Deater

Married Robert Philip Sorber

- Daughter: Isabelle (Married Leroy Butler)
- Son: Claude A. (Married Eunice Oney)
- Daughter: Judy (Married Joe Neilson)
- Daughter: Alice (Married Larry Steltz)
- Daughter: Julie Dee (Married Irv Markle)
  - 5 Children
  - 15 Grandchildren
  - 17 Great Grandchildren
  - 2 Great-Great Grandchildren

Mom is family oriented. She loves her kids and grand kids. She prays for them and remembers their birthdays and anniversaries. She loves going to parties for them.

She has deep roots in her home. She and dad rebuilt, remodeled, landscaped and raised their children in this home, along with several foster children. Her home is always open to guests and overnight visitors.

She is adventurous and has hunted and bagged several deer. She has fished, but does not like catching eels!

When I was five years old, she took me to church with her one Sunday evening. When she got home, dad asked, "Where is Claude?" I was still asleep on the pew at church. With no phones at the time, the pastor waited for her return.

Mom likes her morning coffee and enjoys working the daily crossword puzzle. She may have called you for an answer!

She will play you a good game of scrabble or Phase 10 and is always willing to learn a new game to play.

She faithfully feeds the birds, winter and summer. She can tell you their species without looking them up in her bird books. All of her visitors check to see what birds are visiting her feeder that day.

The squirrels steal the seeds; she grabbed one by the tail and ended up with the end of the squirrel's tail as a souvenir.

The bears also like the seeds, so she would take the feeders in before dark. One bear showed up before dark, and she swatted it with her broom. A bear also broke the window screen and climbed into the mud room where she keeps cans of bird seed.

We had an old Jeep—no top, no sides—that she used to take us kids to the creek for a swim and picnic on a hot summer day. She still can be found by the cool water when it is hot and muggy.

She will be at the Deater Reunion in July, with a smile and a pan of baked beans. Tell her "hello" and give her a hug.

June Deater Sorber

My mother is my best friend, my mentor, my inspiration. She has been my counselor and my advisor. She has comforted me in my grief and uplifted me when I was low.—Submitted by Alice Steltz

DFI

To My Mother

By Julie Wood

My mother hardly ever calls me by my name.
Alice, Judy, Belle, are a few to name.
She knows exactly who I am, but she calls me Boot again.
My mom is not mixed up or confused when she says Belle tie your shoes.
She knows it's me that she is speaking to
But she calls me Beulah very often, too.
I am not worried or upset
To think I remind her of Beulah yet.
I do the same, you know, when I talk to my children, also.
I call them who they are not and they remind me very often yet.
I say, too, I know who you are.
I was just thinking of your sister who moved away so far.
A split personality is what I must have
To be called by so many names in a conversation my mom and I had.
It's not a person she sees,
But the love of God we share from seed to seed.

I love you, mom.
Guess Who

My Hero

By: Autumn Wood

In our world today, everyone has a hero. When I was first asked who my hero was, I said, "I don't have one." Whether the reader knows it or not, everyone has a hero. I would like to tell you about mine. She is June Sorber, my grandmother.

One quality I like about her is the way she is giving. We had a big grease fire in our kitchen once. Everything was black—the curtains, the ceiling, the stove—it was all black. Eight people live in our house, so we are always "going, going, going." She came down to our house and scrubbed everything. My grandmother comes down to our house giving her time and energy, picks up a bucket of soap and water and says, "OK, where do you want me to start?" While she works, she is so happy and sings.

Another great quality I like about my gram is she is very family oriented. She loves family gatherings. Every holiday she calls everyone to get them to come for dinner. We all go to her house or Aunt Judy's house. She goes to every family reunion and reminds us about it. She really loves her family.

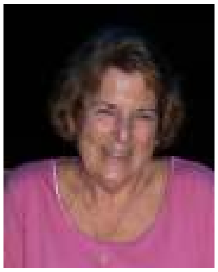
The best quality I think my grandmother has is that she is so full of energy. She is seventy years old and still has a job. Besides having a job, she cleans her house. This house is spotless. This is no small house either. A four bedroom, two bath, living room, dining room, and even attic, basement, and porch. She mows and rakes her yard. She is a Sunday School teacher at her church. Sometimes I wonder who has more energy, she or I.

A hero is a great thing to have or to be. Heroes come in all different ages, shapes, sizes and colors. A hero is someone close to a person or someone a person knows by reading about them. Most of all, a hero is someone to look up to.

DFI

Mutant SPTLC1 dominantly inhibits serine palmitoyltransferase activity in vivo and confers an age-dependent neuropathy.

Submitted By: Ellen Burns, Medical Liaison



The very impressive title is the name of an article published in *Human Molecular Genetics, 2005, Vol. 14, No. 22. pages 3507-3521.*

The primary author of this publication is Alexander McCampbell, who is responsible for the development of the mouse model for HSN-1. Others whose expertise contributed to the report include Dr. Robert H. Brown, Junior, Director of the Day Laboratory for Neuromuscular Research, Charlestown, Massachusetts and Dr. Teresa M. Dunn, Department of Biochemistry and Molecular Biology, Uniformed Services University of the Health Sciences, Bethesda, Maryland.

Mutations in enzymes involved in sphingolipid metabolism cause a variety of neurological disorders, but how this happens, at the cellular level, is not known.

A mutated gene has been associated with HSN-1. It is identified as SPTLC1. Genes make proteins; proteins make enzymes. SPTLC1 encodes one subunit of the serine palmitoyltransferase enzyme (SPT), the rate-limiting enzyme in sphingolipid synthesis. This enzyme is known to affect the production of a fatty substance (glycosyl ceramide) in the body. HSN-1 patients have reduced SPT activity.

As we know, hereditary sensory and autonomic neuropathy type one (HSN-1) is an adult onset, autosomal dominant neuropathy (a parent must have the mutated gene for it to be transmitted to a child, but only one parent must have the gene). HSN-1 patients have sensory and motor loss and often have ulcers, weakness, and poor muscle reflexes. Both myelinated and unmyelinated nerve fibers are affected. The nerves of the dorsal root ganglia of the lumbosacral region (the lower back) are most severely affected.

Earlier experiments involving cell cultures indicated that mutant SPTLC1 inhibits SPT activity. In this study, transgenic mice (mice with a gene added) were created to over express either normal SPTLC1 or mutant SPTLC1. The mice with an added mutant gene developed age-dependent weight loss and mild sensory and motor impairments. Aged mice with the mutant genes lost myelinated nerves in the dorsal and ventral roots of the spinal cord. The mice did not develop skin ulcers on the toes and fingers, but they did develop decreased sensitivity to thermal pain (heat). The overall neurological involvement in the diseased mice appears less severe than the advance stages of the disease in patients.

The mice that have the altered genes are a new mouse model of peripheral neuropathy and confirm the link between mutant SPT and nerve dysfunction. What is still unknown is if the biochemical changes cause the pathology of the disease.

It is noted in the paper that another dominantly inherited, hereditary sensory neuropathy with ulcers, Charcot-Marie-Tooth is also caused by a genetic mutation that disrupts sphingolipid metabolism in another way.

More work needs to be done with the mice to further define the exact mechanism of the disease. Alex has proposed other testing that will continue at the Day Lab to answer some of the questions that have been raised.

The mice that have the altered genes confirm the link between mutant SPT and nerve dysfunction.

Future research may determine if the bio-chemical changes CAUSE the pathology of the disease.

DFI

Update on Khemissa and Paul

By: Ellen Deater Burns

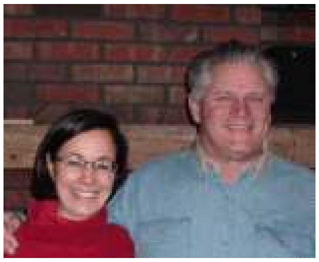
The message on the cell phone was, "I hope you packed your long johns, the temperature last night was 23 below zero." It was February, and we were in Minnesota, a long way in miles and temperature from Florida! The caller was Paul Olinski, and we were on our way to visit him and Khemissa Bejaoui, who live near Marshfield. Khemissa, many of you will recall, was the researcher at the Day Lab who independently identified the genetic defect that causes HSN-1. She took a personal interest in the family and memorized the names and relationships of the members of the family!

In 2003 she left the post-doctoral position through which she did work on another myopathy and then HSN-1 at the Day Lab. She now has her own lab at the Marshfield Clinic. We spent a weekend at their home, set on several acres outside of town. The large "living" room is the original homesteader house, with fireplace, high ceiling, cozy seating, and table and chairs. Out the back window we could easily see the dozen or so deer who came to raid the corn crib, and the flock of wild turkeys who came right to the back door, looking for a hand out!

Khemissa took us to the prestigious Marshfield Clinic, which serves people from Wisconsin and Michigan, and the Marshfield Clinic Research Foundation, which focuses on diseases that impact the lives of Midwesterners.

Dr. Bejaoui's state-of-the-art laboratory is involved in research for Multiple Sclerosis. As part of her work, Khemissa has interviewed many people, some with various neuromuscular diseases. Some of the people she has met are named Dieter. A letter from my Great-Aunt Alice Deater Greevy to my father, Harvey Deater, mentions visiting "Aunt Clara" in Kingston, Pennsylvania. Aunt Clara said that her son, Ivan, had received a letter from a cousin in Wisconsin shortly before he died. Aunt Alice wrote, "*He*" (the cousin) "*said his grandfather and my grandfather came over together from Holland, then my grandfather came East but his gr-f- stayed there and we have loads of relatives there. Maybe that is why my grandfather took his family to Wisconsin when his wife died and was buried.*" We wonder if there is any connection?

Khemissa and Paul have good memories of attending the Deater family reunion, and send greetings to all.



> **The message on the cell phone was, "I hope you packed your long johns, the temperature last night was 23 below zero." — Paul Olinski**

Khemissa and Paul send greetings to all.

Website Comments, Suggestions, Questions, etc.

Contact Dixie Dorward:

ddorward@deaterfoundation.org

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

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DEATER FOUNDATION, INC.

P. O. Box 255
White Deer, PA 17887

DFI

We're on the Web!
www.deaterfoundation.org

Deater Foundation, Inc.

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

An Overview of the Foundation

Submitted July 2005 by: Carol Dorward, President

Since its origin in 1990, funding research to find a treatment and/or a cure for HSN-1 has been the primary goal of Deater Foundation, Inc. In only 15 years, we have witnessed research success with the identification, in 2001, of the diseased gene. Family members can now choose to be tested (via a blood sample) to determine whether or not they carry the defective gene. And now we look forward to research news regarding treatments for complications of the disease and/or pro-active methods of preventing the onset or progress of the disease in affected family members.

Thankfully, over the years, we have had the interest and support of significant individuals in the medical profession—all of whom have been directly responsible for the success of the research which benefits our family. We are grateful for all who have helped us.

We appreciate, too, our own family and friends who have faithfully supported the research with prayer and finances. It is incredible to think that more than $216,000 has been donated to The Foundation over the past 15 years. As a result of the contributions (and interest), The Foundation has been able to support the research with more than $180,000 in that same span of time. Amazing, isn't it?

The research continues; consequently, DFI covets your continued support, both in prayers and in finances.

As the family grows and technology advances, we would do well to "keep up" as best we can in order for future generations to reap the benefits of what treatments/practices work and what doesn't. Therefore, we urge family members who are able to check out the Deater Foundation website [**deaterfoundation.org**] and especially consider joining "The Forum" in order to share questions/comments/suggestions or "what works"/"what doesn't work" [where treatments are concerned], symptoms, etc. The Forum is open **only** to **our family members**. It is a family link all year round, 24/7, instead of the once-a-year reunion.

In the event that you don't have a computer (or are computer shy), consider providing any of the DFI officers with the information you'd like to include in "The Forum" database (perhaps via a written summary…or through a phone call). Then the DFI officer can enter your information in "The Forum" for you.

Be sure to provide your email address to me at (cdorward@deaterfoundation.org) for the DFI email group in order to receive information sent periodically throughout the year. **It is vital that we stay connected.**

Thank you, once again for all you do to insure that the research for treatment of HSN-1 continues.

DFI Treasurer's Report (May 31, 2005, to May 31, 2006)

Submitted by: Nancy Newcomer, Treasurer

DFI Statement of Account

Balance as of 5/31/05	$48,913.30
Income 6/1/05 to 12/31/05	6,980.00
Interest 6/1/05 to 12/31/05	136.22
Income 1/1/06 to 5/31/06	250.00
Interest 1/1/06 to 5/31/056	108.72
June 2005 Expense	- 8,000.00
Balance as of 5/31/06	$48,388.24