

RESEARCH REPORT—2004

SUBMITTED BY ELLEN BURNS, VICE PRESIDENT & MEDICAL LIAISON

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IN MEMORY OF THOMAS DEATER 6

REUNION REMINDER!

Hope to see you at the 60th Deater Reunion on July 17, 2004 at the Grove in the Stull!

The Deater family has suffered for generations with a disabling neurological disease. And yet, courage, humor, faith and determination are the traits that come to mind when thinking about the family. These traits have been the basis for strong relationships that have formed with medical and research people studying the disease.

This past year, Dr. Khemissa Bejaoui, who worked on the HSN1 project at the Day Lab for about 10 years, accepted a new position at the Marshfield Clinic in Marshfield, Wisconsin. One of the first things she did after her arrival was to attend a conference related to HSN 1. The family developed great affection for her on her frequent trips to the reunion. We wish her well and look forward to hearing about her continued research.

Two weeks after last year's reunion, two researchers who had collaborated with Khemissa arrived in Noxen to conduct a research study. Beulah and Elwood Womer opened their home as the "research center" for San Francisco Doctor of Pharmacy, Dr. Walter Holleran and Dermatologist Dr. Joachim Fluhr from the former Democratic Republic of Germany (East Ger-

many). June Sorber hosted the doctors overnight as they gathered information that they presented the same week at an international conference in New England. They speculate that, because the gene defect in HSN 1 is associated with sphingolipids, and sphingolipids are essential for adequate skin barrier function, it is possible that trauma to the skin may play a part in the destruction of nerves. A copy of their presentation will be available at the reunion.

Both researchers expressed appreciation for the cooperation and warm welcome they received from the family. Dr. Fluhr wrote, "I would like to thank you and all members of your *phantastic* family. It was a wonderful experience for me to visit you at this place, to hear about your *lives*, about the fears, the dreams, and sharing some exciting moments together. It was not only about science or being a Dermatologist. It was truly a deep experience learning about other families in a part of the world that is different from our lifestyle."

Dr. Holleran invited me, on October 14, 2003, to visit at his office at



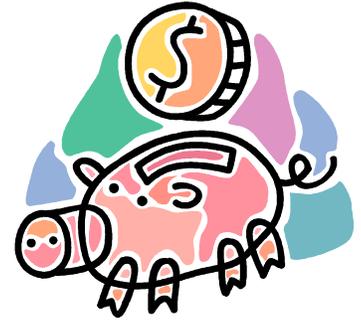
Dr. Holleran and Ellen Burns confer at the "Womer Research Center" August 2003

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TREASURER'S REPORT

SUBMITTED BY NANCY NEWCOMER, TREASURER

2003-2004 Finances	
Balance as of 5/31/2003	\$25,351.67
Income 6/1/2003 to 12/31/2003	13,057.00
Interest 6/1/2003 to 12/31/2003	120.16
Income 1/1/2004 to 5/31/2004	600.00
Interest 1/1/2004 to 5/31/2004	38.39
Balance as of 5/31/2004	\$39,167.22



Contributions to DFI are tax deductible!

Send donations to:
Mrs. Nancy J. Newcomer,
Treasurer
Deater Foundation, Inc.
PO Box 255
White Deer, PA 17887

GRANDMA'S DIARY — CHRISTMAS AT THE OLE HOMESTEAD

SUBMITTED BY BEALUH WOMER AND LAURA MAE DEATER

Thursday, December 24, 1942—

WARMER Well today it was warmer we baked mince pie & apple tart bread & cinnamon buns stuffed a turkey after making stuffing washed windows made cranberry sauce mopped the floor washed lamps & windows the girls washed their heads there was a lot of water carried we did a lot then trimmed the tree just as we had trimmed the top put the tinsel on Lynn & Isabel came They helped finish it then Lynn Isabel Harvey Helen & Verna went to Tucksville for Russell Then June and Bob came Martha & Beulah were to bed Mona & Carl

had come before June & Bob and we had laid out all the presents oranges candy & nuts Little Isabel had quite a time over the tree & the dolls when June got ready to go homeshe discovered she had lost her purse she went back to Spencers to see if it was there but it wasn't It was late when the bunch got back from TRucksville and Lynn Isabel & Russell hadn't had supper yet so I fried eggs and made sandwiches coffee and cookies of course the other ones came for cookies & coffee too well Isabel laid out the other presents and after the regular visit we went to bed that was about 2 o'clock

Friday, December 25, 1942—

NOT SO COLD Well the big day everybody has opened their presents and sure filled up at the dinner table the men are playing cards some of the other carroms the girls running over to look at Keiper's presents as they are having an extra nice Christmas at night Tom Lois Janet Tommie Jr & Tommie W. Russell had gone to bed Tom & Tommie Wilson went up to see him after visiting a while they went down to Mona's for the night but Janet ray & Tommie stayed here June and Bob went home. Martha went with them



DR. WALT HOLLERAN

SUBMITTED BY ELLEN BURNS

Dr. Walt Holleran is a research professor and an expert in the metabolism of specific lipids present in the skin and other tissues, called “sphingolipids” or “ceramides.” He has been studying the function of the skin as a barrier, and the role of ceramides in forming the barrier in the outermost layers of the skin, called the stratum corneum. Concentrated and organized layers of lipids, including ceramides, are found between cells in the stratum corneum.

This combination of “dead” skin cells and surrounding lipids constitutes the primary barrier between humans and their environment.

As you are aware, recent work by Drs. Bejaoui and Brown revealed that specific gene alterations in affected family members include changes in the gene for an enzyme in ceramide production, called serine palmitoyl-transferase (or “SPT” for short). Dr. Holleran, who has conducted nu-

merous studies into the role of SPT in normal skin function, was contacted by Dr. Bejaoui to help determine whether the skin might also be altered in affected individuals.

To this end, Dr. Holleran and a colleague, Dr. Joachim Fluhr, visited last summer with a number of family members to measure specific parameters of skin structure and function. Their initial results show that

(Continued on page 5)



Dr. Walter Holleran

“Dr. Holleran, was contacted by Dr. Bejaoui to help determine whether the skin might also be altered in affected individuals.”

THE “DEATER DISEASE” — A HISTORICAL RESEARCH REVIEW

SUBMITTED BY ELLEN BURNS

Hereditary Sensory Neuropathy Type I (HSN I) is a rare genetic disorder characterized by the loss of sensation, especially in the feet and legs, and progressing to the hands and forearms. The loss of sensation is caused by abnormal function of the autonomic nervous system, which controls responses to pain and temperature as well as other involuntary or automatic body processes.

The most common type of inherited sensory neuropathy is Charcot-Marie-Tooth (CMT) disease. Some texts do not differentiate between this more common neuropathy and any other sensory neuropathy. Doctors who are not specialists in this area may diagnose someone with HSN I as having CMT.

In 1922 E. P. Hicks published an article in an English medical journal describing

an English family in which 10 members suffered from perforating ulcers of the feet, shooting pains and deafness. In 1951 D. Denny-Brown published a follow up report on one of the members of this family, identifying the disease as “hereditary sensory radicular neuropathy.” In 1939 Drs. Tocantines

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Drs. Holleran and Fluhr with Beulah and Elwood Womer

Aug 2003

RESEARCH 2004 [CONTINUED]

the VA Hospital in San Francisco. He provided a supply of TriCeram cream, for use by affected family members. I sent it to those involved in the study and hope to get a report on any improvement anyone may have noted after using the cream. Dr. Holleran wrote recently that his son will attend Bucknell University in Lewisburg PA starting this fall, and he hopes to visit the family again. Dr. Holleran will be attending two conferences in Japan in July to share his findings with other researchers.

Dr. Robert Brown has been involved with the family for over 20 years. As Director of the Day Laboratory for Neuromuscular Research, he has directed ongoing research on HSN1.

He was recently awarded a grant from the National Institutes of Health for a collaborative research project on HSN1 with Dr. Theresa Dunn and colleagues at the Uniformed Services Medical School in Bethesda, Maryland. This is an especially exciting collaboration for us. In June, 2001, Dr. Todd Martensen, who worked with Dr. Dunn, contacted the Deater Foundation web site, which was developed and is maintained by Dixie Dorward. I responded to his inquiry and put him in touch with Dr. Brown and now both groups are working together on exciting new research!

Dr. Brown reports "The work has been productive over the last several months." He has visited

Dr. Dunn twice for scientific exchanges and reports "she is an excellent scientist who is knowledgeable, effective in communication and a pleasure to work with." At the Day Lab, the grant is fully funding the work of Alex McCampbell's postdoctoral studies. Alex is skilled at developing transgenic mice and Dr. Brown reports "we are well on our way to



August 2003 "Womer Research Center" Participants

develop a mouse model."

The development of a mouse with the same genetic defect as people with HSN1 would allow for in depth study of the disease. The defective gene must be inserted into the test mice. Then the mice must be shown to pass the defective gene on to offspring. Once the genetic defect has been confirmed, the mice will be carefully studied for any symptoms of the disease. Dr. Clifford Wolf, who has offices in Charlestown near the Day Lab, is a pain specialist who is expert in pain testing in mice. He will be a resource for the studies. Other tests will include testing the skin of the

animals for small fibers of nerves. When (and if) a true HSN1 model mouse is developed, the mice would be made available to other researchers for continued studies.

The research team has a parallel goal of developing a miniaturized system to study the gene defect in the laboratory in Petri dishes. Drugs can be placed in conjunction with the "test tube" disease. This research would eventually allow for the screening of many drugs simultaneously. The long-term goal will be to discover a treatment for the disease!

Dr. Brown's older daughter, Hannah will start medical school at Harvard this fall. His younger daughter, Rachel, is at Brown University. It was fun to find out that Dr. Brown's wife, Elaine, used to vacation with her family in Maine, when she was a child. They often ate at "Spillers" restaurant- that is the same building where Larry Deater's restaurant "The Privateer" is today!

God works in wondrous, mysterious ways! He has taken the prayers and financial contributions we have offered and has expanded them in ways we could not have dreamed! The gifts of money made to the Deater Foundation are like grains of mustard seed. When watered with faith, they grow by stimulating other contributions, and the studies they support encourage scientists in research that spans the globe!

DR. WALT HOLLERAN (CONT'D)

SUBMITTED BY ELLEN BURNS

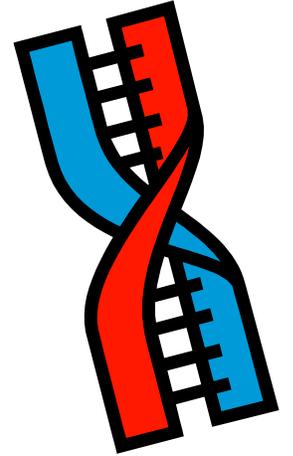
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skin of affected family members may have a very slight delay in the ability to recover from a mild challenge; that is, the outermost layers of dead skin cells and surrounding lipids were removed using sequential applications of tape. However, initial microscopy studies have so far

revealed very little difference in the structure of the stratum corneum in affected vs. non-affected individuals.

Although these preliminary results suggest that the outermost layers of skin may be altered in affected individuals, it must be stressed that the relationship of these find-

ings to the progression of symptoms will require further study and analysis. Dr. Holleran hopes to continue these studies in conjunction with Drs. Bejaoui and Brown, and to determine whether skin structure and function relate to progression of symptoms in affected family members.



“Dr. Bejaoui found that the mutations in the gene resulted in reduced enzyme activity and sphingolipid production. This discovery moved the research much closer to identifying what causes nerve destruction and ultimately, to treatment.”

HISTORICAL RESEARCH REVIEW (CONT'D)

SUBMITTED BY ELLEN BURNS

and Reimann published their first report on Harvey and Russell Deater and other members of the family. Drs. Reimann, McKechnie, and Stanisavljevic published a follow up report in 1968. Similar symptoms were noted in French-Canadian, English, and Australian families.

During these early years, neurological exams, biopsies, and nerve conduction studies were the only means of identifying the disease. There was some variation in the symptoms, including restless legs, severe sensory loss, and burning feet. Various members of the Deater family participated in studies at The

National Institutes of Health during the 1970s with Dr. John Whitaker and Dr. Marinou Dalakas.

In 1986 Dr. Robert Brown drew blood from family members to begin a study at the Day Neuromuscular Lab at Massachusetts General Hospital. By 1992, Dr. Khemissa Bejaoui was involved in the research. At the turn of the 21st century the method of diagnosis changed when Dr. Garth Nicholson in Australia identified a genetic mutation causing the disease. Very soon after that Dr. Khemissa Bejaoui independently identified mutations in other patients, including those in the Deater

family.

The mutations on chromosome 9 were slightly different in different families. The gene involved is a known gene, SPTLC1. Many people around the world had already been studying this gene and the enzyme associated with it. Dr. Bejaoui found that the mutations in the gene resulted in reduced enzyme activity and sphingolipid production. This discovery moved the research concerning HSN 1 much closer to the goal of identifying exactly what causes nerve destruction and ultimately, to treatment.



Denise Gordon and Connie Fleeger

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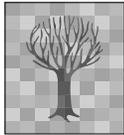
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IN MEMORY OF THOMAS DEATER

- 5th Child of Alvin and Ellen (Wilson) Deater
- June 6, 1915 to July 7, 1970
- Married Lois Babcock
 - ◊ Son: Tommy Deater
 - ◊ Son: Ray Deater
- Married Frances Hickox
 - ◊ Stepdaughter: Sally (Spear) Rood
 - ◊ Daughter: Tonja (Deater) Cooper-Harris
- 10 Grandchildren
- 6 Great-grandchildren

Dad always loved the outdoors and cherished the time he was able to hunt and fish. He loved and valued his family; I never once doubted this. His bond with his brothers and sisters was ingrained in his nature, just as it is with all his brothers and sister. No greater legacy could be passed down through the Deaters than this bond that touches us all. — **Tonja (Deater) Harris**

