

Research 2003

NBIA Disorders Association is sponsoring two \$30,000 seed grants through the National Organization for Rare Disorders in 2003. As you know, three grants were made available in 2002. Those research projects are now underway.

Thanks to all who sent in donations in December to help fund the two new grants. We fell short of our goal of \$10,000, but the board voted unanimously to take money from the General Fund so we would not miss this opportunity. We still have adequate resources to handle our expenses. Notices have gone out to hospitals, universities, researchers and medical journals asking for abstracts by March 15, 2003. It was also a wonderful opportunity to make everyone aware of the new name of the disease and our organization at no additional mailing costs.

These research grants are vital to encouraging new researchers to study our disease. In February, NBIA Disorders Association was contacted by a researcher who received the RFP (Request for Proposals) notice sent out. He decided our disease was one he would like to study and plans to apply for one of the grants.

This is how our organization can make a difference and help research move forward more rapidly.

Please continue to support our Research Fund through your donations and fundraising efforts.

Inside This Issue...

Sinemet Survey	2
Research Update	4
NBIA in Gerny	5
Memories	6
Our Generous Supporters	7
Message from the President	7



Dietmar Klucken, 12, with friends.

Coping with NBIA/PKAN in Germany

By Angelika Klucken

Our first son, Dietmar, was about 8 years old when he developed the first distinctive symptoms of dystonia and dysarthria (rigidity in muscles) shortly after a chickenpox infection. He was 11 when he was diagnosed with HSS, now known as NBIA, in October 2001.

After that frightening diagnosis, we left the University Hospital without any hope. All of the neurologists we consulted were at a complete loss. Then, we heard of your association and drew new hope from reading the Web site. Everyone we contacted in the U.S. for advice in the following weeks was friendly and promptly responded to our desperate questions, thus giving us comfort and helping us to get through the months after the diagnosis.

In spring 2002 genetic testing revealed that Dietmar has PKAN. He refused to go out for walks because generalized torsion

(see Dietmar on p.3)

HSSA is now NBIA Disorders Association

After much discussion and planning, Hallervorden-Spatz Syndrome Association is now the NBIA Disorders Association. The name change took effect in January once all of the official paperwork was filed. The association sought the name change as the disease was renamed due to the unethical activities of Hallervorden and Spatz during WWII. The moniker embraces the new name for the disease, which is being called neurodegeneration with brain iron accumulation or NBIA. The Web site also has been updated and revamped to reflect changes. Check it out at www.NBIAdisorders.org.

MEDICAL ADVISORY BOARD

Susan Hayflick, M.D.

Geneticist & Pediatrician
Oregon Health & Science University
Portland, Oregon

Rayburn Skoglund, M.D.

Pediatric Neurologist
Childrens Associated Medical Group
San Diego, California

Kenneth Swaiman, M.D.

Pediatric Neurologist
University of Minnesota
Minneapolis, Minnesota

Ramesh Tripathi, M.D., Ph.D.

Dept. of Ophthalmology
University of South Carolina
Vision Research Laboratories
Columbia, South Carolina

Officers/Board of Trustees

President - Patricia V. Wood

El Cajon, California

Secretary - Debbie Forstall

Northridge, California

Treasurer - Gayle McMahon

El Cajon, California

Board

Dianne Gray
Naples, Florida

Susan Laupola
Cincinnati, Ohio

Mary Ann Roser
Austin, Texas

Kris McGourthy
Middleboro, Massachusetts

Mary Tapke
Cincinnati, Ohio

Disclaimer

The views expressed in the NBIA Disorders Association newsletter do not necessarily represent the views of the Board of Trustees or the Medical Advisory Board. Check with your doctor before trying anything new.

Association surveys families on drug benefits

The NBIA Disorders Association mounted an e-mail survey in January after a family requested information about Sinemet, a drug normally used to treat people with Parkinson's disease for tremors. The association is grateful to the families who responded and is sharing the results, which are not scientific but could be helpful when discussing approaches with patients' doctors.

Of the families who responded, eight NBIA patients, including two girls in the same family, reported no improvement from taking Sinemet for relief of any NBIA symptoms. Those patients stopped taking the drug.

In three other patients, improvements were reported. Of those, one said the drug helps but that when too much is given, the result is "dancing legs," (involuntary movement of the legs). Another said the Sinemet helps muscle spasms best when given with Baclofen. The third patient, who has been on the drug for a year, said it helps with tremors and dystonia. He takes 25/100 mg 3 times a day and said it works best during the day when he is awake.

Another patient in the survey became so heavily sedated by the drug he stopped taking it. That patient, however, may try it again. Another felt there was a decrease in muscle spasms and that it helped, but the side effects of dyskinesia and nausea were overwhelming and outweighed the benefits. The drug was stopped after two weeks.

Physician Referral Network

Letters have gone out to NBIA families requesting information on their physicians who deal with NBIA. We are building a database for referral purposes so that new families will have somewhere to turn when looking for a doctor familiar with the disease. We also plan to build a network for the physicians, so that they can share experiences and therapies they have tried and help expand the knowledge base about NBIA.

If you are a NBIA family and have not yet received a letter and would like to participate with your physician information, please contact us at info@NBIAdisorders.org and we will add you to our NBIA families database.

Please be sure to take the time to fill this form out and return it to us promptly. You could make a real difference in another family's struggle with this disease.

Dietmar

(continued from pg. 1)

dystonia disabled him increasingly. After an unsuccessful trial with L-Dopa/Carbidopa, Dietmar began to take vitamin B₅. In June, he was up to 2000 mg daily and his complaints about painful muscle cramps in his left foot vanished. He began to feel better.

During June and July of last year, we tried other treatments as well. Dietmar continued the B₅ therapy but also took a course of medical treatment at a neurological Reha-Hospital for children. I accompanied him during those six weeks. The pediatric neurologist recommended Artane (Trihexyphenidyl), and, very slowly, we increased the dose from 2 mg daily to 15 mg. At about 8 mg Dietmar's speech markedly improved. He perspired less and the dystonia in his back seemed to get a bit better. But the higher the Artane dose, the more side-effects: restlessness, dyskinesias, cotton mouth and dry skin. So we decided to reduce Artane slowly until we got down to nothing. We wanted to find out if Artane really had any positive effects. Two weeks after we had taken Dietmar off Artane, he was bathed in sweat again every day, had trouble speaking and worsening dystonia in his back. At present, he takes 8 mg Artane daily, and we think it really helps him.

During his stay at the Reha-Hospital, Dietmar received a wheelchair we had ordered months previously. The hospital staff saw that it was totally inadequate for Dietmar's size and symptoms. They helped us get a proper wheelchair with a seat that can be tilted backwards to relax his back.

Dietmar also has benefited from other treatments at the hospital:

Physical therapy, including stretching and wheelchair training

Occupational therapy, such as practicing Activities of Daily Life (ADL) like dressing, making breakfast, etc., training fine motor skills and attention

Speech therapy, including training his slightly impaired tongue by speaking with a cork in his mouth

Dietmar also uses the hospital's swimming pool and loves moving in the warm water. He enjoyed riding a special tricycle for disabled children, and the hospital staff helped us get one for Dietmar to use at home so he could train his leg muscles. The recommendations of the hospital staff made it considerably easier for us to get all of the prescriptions for therapies and aid equipment that Dietmar needs. Also, my son and I enjoyed the relaxation and time we spent there with each other. A psychologist was on hand for the parents and she helped me clear my head and comforted my heart.

Since those weeks at the hospital, Dietmar's condition has stabilized, and he has even got a bit better. People who saw him in spring and then in fall were astonished at the turnaround. Two times a week Dietmar gets physical and occupational therapy. He bounces back more quickly from infections and other setbacks now.

Right now, he takes 4000 mg B₅, 8 mg Artane, 100 mg Coenzyme Q10 and 30 mg Baclofen daily. Every two days, he takes a multivitamin capsule. We haven't seen any positive changes from Baclofen so far and aren't sure why. We hope that the B₅ is still working and the other vitamins are contributing.

None of the treatments we have tried have resulted in a miracle, but we have seen the progression of NBIA slow down considerably for the first time in two years. We still put our greatest hopes on the B₅ and hope that perhaps there could be residual enzyme activity in his *PANK2* gene.

The main thing is that Dietmar is happy. He enjoys life, though he is sometimes sad. Most days, he doesn't allow NBIA to keep him down. It's really admirable how fast he has accepted his new life as a disabled child during the last two years. Dietmar has got a special sense of humor and an infectious laugh. He's very creative and persistent about reaching his goals. He's a fan of Eminem, Busted and Linkin'Park. He likes to surf the Internet and to design little letters to his friends with "Print Art Artist." His favorite films are "8 mile" and "Harry Potter."

We're trying to live a normal life—as much as possible. And there is Dietmar's younger brother Matthias, 11, who isn't affected by NBIA but who also needs his family. Our lives are full.

... he has accepted his new life as a disabled child during the last two years. Dietmar has got a special sense of humor and an infectious laugh. He's very creative and persistent about reaching his goals.



Dietmar, Angelika, Matthias and Stephan Klucken from Velbert, Germany

New PKAN study underway at OHSU

By Allison Gregory

A new study of individuals with pantothenate kinase-associated neurodegeneration, or PKAN, is about to get underway at the Oregon Health & Science University. It will be led by Drs. Susan Hayflick and Penny Hogarth, who each were awarded grants of \$30,000 last year from the NBIA Disorders Association.

The association made three such awards last year, its first ever, and will make two more \$30,000 awards later this year. Hogarth and Hayflick are combining their awards to collaborate on this new study, which will investigate the specific features of PKAN and how the condition progresses over time.

About 25 patients of all ages, in both early and late stages of PKAN, will travel to OHSU in Portland, Ore., for a three-day visit to give the researchers a chance to do a variety of assessments on them. The patients and their travel companion will be accommodated at a research support center at the hospital.

Although anyone with PKAN knows from experience that the disease features progressive movement disorders and involves several body systems, the researchers want to study PKAN systematically. By assessing 25 patients in different stages, a large amount of data will be gathered to better characterize the signs and symptoms of PKAN and how it progresses over time. The knowledge gained from this work will lay the foundation for future research, particularly when clinical trials begin for testing possible treatments. Now that the causative gene, *PANK2*, has been found, and the biochemical basis of PKAN is better understood, it is hoped that the OHSU studies may also point the researchers toward potential treatments.

The researchers will use a sophisticated type of MRI with spectroscopy, which assesses the brain for various levels of chemicals. An electroretinogram, or ERG, will also be performed to look for changes in the photoreceptors in the back of the eye. Although many individuals with PKAN eventually develop retinal abnormalities, it is suspected that an even higher number may have changes detectable by ERG.

Also during the three-day visit, a diet assessment, physical and neurological examinations, and blood and urine studies will be done. Hayflick and Hogarth plan to use several standardized testing tools, such as a dystonia scale, quality of life scale and intelligence testing to look at the many aspects of living with PKAN and how it affects an individual's life and function over time.

Patients will begin coming to OHSU for the testing within the next few months. OHSU genetic counselors Jason Coryell and Allison Gregory are contacting eligible families about the study. Interested families may also contact them for more information at (503) 494-4344 or to coryellj@ohsu.edu or gregorya@ohsu.edu.

Mouse model in progress

Dr. Han-Xiang Deng of Northwestern University Medical School in Chicago, Ill., was the recipient of our third research grant in 2002. He says his lab has begun the work of generating a NBIA mouse model — a process that generally takes approximately one year to complete. Dr. Deng will keep us informed as to the progress of this project.

New genetic counselor joins Oregon university staff



Allison Gregory

Dr. Susan Hayflick's NBIA research team at the Oregon Health & Sciences University in Portland, Ore., has a new genetic counselor to work with families who have a NBIA diagnosis: Allison Gregory. She will work part-time and joins genetic counselor Jason Coryell, who also is on the team and attending medical school.

Gregory's work has been in prenatal diagnostic settings, most recently at Cedars-Sinai Medical Center in Los Angeles. Working as a genetic counselor in a research setting is a new and interesting challenge for her, she said. She will help coordinate family involvement in research studies and serve as a contact person and resource for families and the medical community. She is also involved in the design and implementation of a new study of a small group of individuals diagnosed with PKAN, a form of NBIA.

Gregory graduated from Stanford University in 1994 with a degree in human biology. She earned a master's degree in genetic counseling at the Medical College of Virginia. She said she is thrilled to be back in her hometown with her husband. When not working, she enjoys playing competitive tennis, reading, and spending time with her family and two cats.

Gregory can be reached at OHSU by phone at (503) 484-4344 or by e-mail at gregorya@ohsu.edu. Because of her part-time schedule, she may not be available immediately but should be able to respond to inquiries within a week.

German NBIA families are uniting with new organization

By Angelika Klucken

As it turns out, last year's family conference in Indianapolis was a life-changing event for me. I was overwhelmed to meet families from all over the world united by the same destiny and to meet the team from the Oregon Health & Science University. I was deeply impressed that the team was on hand during the whole conference and gave us so much information about NBIA. Before the conference, I had the vague idea that something had to be done for those NBIA families in Germany who couldn't understand the English material written about the disease. I thought we should unite. At the time of the conference, I already had been in contact with a few NBIA German families and I shared with them my experiences in Indianapolis.

When I returned to Germany, I knew what I had to do. My husband and I began laying the groundwork to establish a German non-profit organization. We called it "Hoffnungsbaum e.V. Verein zur Unterstützung der Erforschung und Behandlung von NBIA (vormals Hallervorden-Spatz Syndrom)." In English, it means: "Tree of hope. Association for supporting research and treatment of NBIA (formerly Hallervorden-Spatz-Syndrome)."

On Nov. 24, we officially launched the organization with three NBIA families, friends, relatives and a young neurologist from the Technical University of Munich where genetic testing for PKAN is done. Our organization's goals are these:

- Support international research and communication between researchers
- Help build a team of specialists to work on NBIA in Germany
- Support early recognition of the disease to avoid misdiagnosis
- Share information and experiences among NBIA families and provide emotional support
- Educate affected families on all questions concerning the disease
- Cooperate closely with the NBIA Disorders Association and support its work
- Make information about NBIA public wherever it could be useful to get support for our work.

For now, we're writing articles for medical magazines, searching for more NBIA families in Germany, translating information about the disease into German and planning fundraisers.



November 24, 2002, the first meeting of NBIA/Germany non-profit held. Pictured is — Julia Zell (15), Tuba Aydin (10), and Dietmar Klucken (12); second row: Uschi Zell-Reichardt, Angelika Klucken and Hatize Aydin.

The NBIA Disorders Association taught us that we really can do something to help our kids. If we've got hope, our children will have hope too. If we're strong, our children will be strong too. If we enjoy life, our children will enjoy life too. By helping each other, we're helping ourselves and our children.

We're very grateful to you.

(For more information, see their Web site at www.hoffnungsbaum.de)

Fundraiser in Claremore, OK

The Mayfields of Claremore, Okla. — Scott, Rhonda and daughter, Kelsey, who has NBIA/PKAN — are doing their part to help the NBIA Disorders Association reach an important goal: raise money for the Research Fund.

The family plans to hold a dinner and magic show April 26 at the First United Methodist Church, the church they attend in Claremore. They will sell tickets for \$10 each and take donations. They will also be selling Home & Garden Candles for \$8 each, and they will make \$4 on each one.

Kelsey will have her picture taken at a studio with the magician and his dove for the flyers and advertising. Also, an article is planned for the Claremore Progress & Oologah Leader papers. "We have had a tremendous amount of support from friends, family, neighbors and church members," Rhonda said. "We are looking forward to raising money for research."

Please contact info@NBIAdisorders if you are interested in having a fundraiser. We have handouts and information on fundraising and will be happy to assist you with your planning.

Susie Miller

Oct. 12, 1986 – Dec. 2, 2002

Susie Miller, who helped inspire Dr. Susan Hayflick to study NBIA, has died at age 16. Susie was born Oct. 12, 1986, and lived in Clymer, N.Y., with her parents David and Mary Miller, and her sisters Amanda, Amy and Anna. She died Dec. 2 at home.

Susie, Amanda and Amy were diagnosed with NBIA (HSS) in 1989. Anna does not have the disease. The Miller family and the Amish community that has several other affected individuals, was Hayflick's first experience with this disease and prompted her to focus her research on NBIA.

Susie attended Hewes Educational Center in Ashville, N.Y.

You can honor the memory of a loved one or a friend through a gift to NBIA Disorders Association. The thoughtful people listed below have made a donation on behalf of their friends and loved ones during the last few months.

In Honor Of

Jim McNulty

Jane M. Roberts

Ashley Middendorf

Memorial Rehabilitation Services

In Memory Of

Amy Hall
Bill & Yvonne Hall

Richie Roberts
Bernie & Judy Roberts

Susie Miller
Hewes Ceter
David & Mary Miller
Gregory & Sarah Parmarter

Clarence Roser
Ted Thomas & Mary Ann Roser

Tony P. Venuto
Bernie & Judy Roberts
Ted Thomas & Mary Ann Roser

Memories of Susie

By Mary Miller

When I think of Susie, I think of the way she smiled and made her eyes sparkle, and how she occasionally showed her temper when we misunderstood.

It was wonderful when she showed other people how much she understood.

It was sad when we couldn't get a smile from her because she was so miserable.

She was one of the sweetest people I ever knew.

I remember how much she talked and how fast she walked between ages two to four, as if she knew she didn't have much time.

What Susie liked best was when her Dad came in to have coffee with her and told her he loved no one else as much as he loved her.

I want to always remember the good times we had with Susie when she was healthy enough to enjoy life.

I'm glad that she didn't have to suffer any more and that she didn't have to go to the hospital or make the decision to have another surgery or not.

There is nothing to say. It is harder than I thought it would be.

I hope the people at school know how much Susie loved them and that there was no other place she would rather be.

It meant so much to Susie when people took time to talk to her, hug her, or kiss her on the cheek.

Our family is sad without Susie here with us, but we try to be happy for Susie and we like to think of her as an angel that can fly, run, talk, eat and play with all the other angels that have gone before her.

MESSAGE FROM THE PRESIDENT

A New Beginning



Patty Wood

The new year began with a bang: a new name, a new Web site, a new logo, a new newsletter format and ambitious plans for the rest of 2003.

A major goal is to support fundraisers organized by NBIA families so we can continue awarding research grants in 2004 — and beyond. We ask that families really take some time this year to think of ways they can help raise money for research. It takes more than just a handful of families to meet this critically important goal. After all, each of us wants a cure for NBIA, and research is our best hope. That, of course, takes money.

Another key goal is to establish a physician referral base for doctors and families to use. This would help new NBIA families locate physicians in their area who are familiar with the disease and its unique characteristics. It also would connect physicians with each other so they could share experiences and expertise. We hope to have this valuable tool available at our Web site.

Once this is in place, our plan is to expand the database so that other specialists, such as physical therapists, speech therapists, occupational therapists, teachers and home care providers, can share ideas about meeting the needs of NBIA patients.

Near the end of the year, we will gauge interest in holding a third family conference in 2004. If families are interested, we will ask for suggestions on topics to discuss at the conference and where it should be held.

We also want your feedback on the new Web site. Are there ways it can be more helpful to you? Is it easy enough to navigate? Is anything important missing? To help you find information quickly, we have added a site search function where key words can be typed in and you will be led to wherever they appear on the site. We would also like to add more pictures to our NBIA families page. If you have previously sent in a picture and would like to update it, please send the new photo (up to two per family are permitted) as an attachment via e-mail or a hard copy via postal mail. NBIA families who have not been represented are invited to add their families to this portion of the Web site.

We now have 2000 people on our mailing list, so the newsletters will be printed professionally. The job was getting too big for my printer and I could better spend the time on more important work, such as helping the organization grow.

Your input is always welcome on these matters and any others you want to discuss. My new e-mail is pwood@NBIAdisorders.org so please be sure to update your files.

I hope you like our "new beginning" and share my excitement for the new discoveries on the horizon for NBIA.

NBIA Disorders Association is grateful to its supporters for their generosity. We extend our deepest thanks to the contributors listed below who have donated in the past few months.

Sandra Beland	June McClure
Barbara Belcher	Lori Mitchell
Bruce Belcher	Mr. & Mrs. Douglas Mulhair
Jeff Bisnaw	Charles & Diane Murdock
Stanley Bryant	Regina Neal
Slava Buchkovich	Linda Grundstrom Park
Pete Cassidy	Lisa Patton
John Collins	April Penner
Dectron, Inc.	Picchio Pharma, Inc.
Mr. & Mrs. Roger Denson	Gloria Rillo
Diageo North America	Anella Roser
Charles Greene	Mr. & Mrs. Carmen Roser
Lee Hamer Memorial Charities	Mr. & Mrs. Andrew Shetler
Jim Hayes	Dr. & Mrs. Rayburn Skoglund
Marion Hermann	Maxine Smyth
Mr. & Mrs. George Jones	Mr. & Mrs. Robert Starling
Mr. & Mrs. Lowell Judd	Jean Steck
William Katz	Ralph E. Steck
William Kelso	Al & Pam Stromsta
Angelika Klucken	Carole Thorp
Cheryl Lamos	Paul & Mary Tierney
Barbara Levinson	Edith Vedova
Richard Mann	Dan Venuto Construction
Jennifer Marik	Mr. & Mrs. Rod Walker
Paula Rodrigues Martins	Raymond Wong
Sebastiao Martins	

Thanks to our logo designers

Our organization is grateful to Dean and Lori Mitchell who donated their time and talent to design our new logo. Dean is a graphic designer by trade, and Lori is an artist and illustrator. We appreciate their help in giving our organization its new look.

Lori has also written and illustrated a children's book that deals with the ways individuals are all different, and yet also are alike. It was inspired by her daughter, April, who has vitiligo, which is a loss of pigment or color in the skin.

You can visit Lori's Web site at www.differentjustlikeme.cc.



2082 Monaco Ct.
El Cajon, CA 92019-4235

Formerly
Hallervorden-Spatz
Syndrome
Association



Our Mission:

NBIA Disorders Association is a non-profit organization dedicated to providing emotional support to families affected by NBIA, educating the public about this disease, and monitoring and supporting research and informing others of its progress.

NBIA Disorders Association

2082 Monaco Ct.

El Cajon, CA 92019-4235

E-Mail: info@NBIAdisorders.org

phone: (619) 588-2315 fax: (619) 588-4093

Visit our Web site at

www.NBIAdisorders.org

from discovery to cure

Formerly Hallervorden-Spatz Syndrome Association