

NBIA sister organization off to strong start in Italy

By Patricia Wood

The NBIA Disorders Association has another European sister. This one's Italian.

Like the non-profit created several years ago in Germany, NBIA families in Italy came together last year to form the Italian Association for Neurodegenerative Syndromes with Iron Accumulation, or as it is known in Italian, the Associazione Italiana Sindromi Neurodegenerativi da Accumulo di Ferro, or AISNAF.

It started when NBIA mother Anna Focaccio wrote to Telethon, a private, non-profit in Italy that helps fund research for rare disorders. Telethon posted Focaccio's letter on its bulletin board and when other NBIA families responded, helped organize a first meeting. One of those families was Nicola and Anna Pascale who attended our third family conference in May 2005. Their daughter Valentina has NBIA.

AISNAF was recognized as a non-profit by the Italian government in March 2006. Twelve NBIA families have joined the association, and six others have recently made contact.



Members of AISNAF at their first meeting in Rome, Italy in April 2007.

(see AISNAF on pg. 3)

Volunteers step up to head NBIA programs

By Patricia Wood

In the December newsletter we asked for volunteers to help us bring new programs to the organization, and you responded. We are happy to announce that four program director positions have been created.

These volunteers will be responsible for the day-to-day running of new programs for NBIA-affected individuals and families. The directors will report to the Board of Trustees on their progress.

Michael Cohn of St. Louis Park, Minn., will be director of NBIA Adult Programs. He will address issues facing adults with NBIA, including helping them work with assistive technology, network with others and become self-advocates. He also wants to help adults with housing opportunities, employment, volunteer opportunities, transportation needs as well as educate them on accessibility rights and medications.

He hopes to set up a fun activity for NBIA adults at the next family conference and organize a session on family interaction for adults.

Cohn started a non-profit, "Promote Awareness," to make a difference in the lives of those with disabilities. He has written, produced and marketed diversity-training videotapes to educational institutions, health care facilities and corporate entities, as well as conducted diversity-training seminars.

"There are many NBIA adults who might be in different stages of life and we need to address all stages and the issues they bring," he said.

(see directors on pg. 4)

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What is NBIA?

Neurodegeneration with Brain Iron Accumulation (NBIA) is a rare, inherited, neurological disorder.

The common feature among all individuals with NBIA is iron accumulation in the brain, along with the progressive movement disorder. Patients can plateau for long periods of time and then rapidly deteriorate. The most common symptom is involuntary muscle cramping, called dystonia.

Symptoms vary greatly from one person to the next, partly because the gene affecting them can differ. Different mutations within a gene also can cause a more or less severe form of the disease.

The movement disorders can result in clumsiness, difficulty controlling the body and speech problems. Also common is a degeneration of the retina, which causes night blindness and a loss of peripheral vision.

Some individuals eventually lose the ability to walk, talk or chew food and become totally dependent on others for all their needs.

Our sister non-profits in Germany and Italy who work with us in the promotion of research and treatment of NBIA, can be contacted at the following addresses:

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Disclaimer

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

AISNAF

(continued from page 1)

The all-volunteer organization is led by Dr. Natale Antonio Scalise who has a 10 year old daughter, Francesca, with PKAN. The vice president, Lorella Faltiba, has NBIA, and the secretary is Ugo Scalise, Dr. Scalise's brother. He is a lawyer and cares for all the legal aspects of the organization. Because of his work with AISNAF, he was appointed as a board member of UNIAMO, the Italian National Federation of Rare Disorders and as a member of the Consulting Board for Rare Diseases at the National Ministry of Health.

AISNAF has a nine-member scientific committee made up of doctors, researchers and geneticists who guide the organization on research and medical information, similar to our Scientific & Medical Advisory Board. In addition, five consultants offer their expertise, including Dr. Susan Hayflick, the director of our advisory board.

The consultants who make up the AISNAF Scientific Committee met for the first time last June and plan to identify worthy NBIA research projects that could be funded by Telethon. The committee's coordinator is Dr. Nardo Nardocci, director of the pediatric neuropsychiatry operating unit at IRCCS Foundation of the Neurological Institute "C.Besta" in Milan, Italy. Nardocci attended our First Scientific Workshop held in Bethesda, Md., in 2000. He is a clinical neurologist specializing in movement disorders and along with fellow scientific committee member Dr. Enrico Bertini of Rome, they have as patients all known NBIA individuals in Italy.

Nardocci is planning a clinical trial for the iron chelation drug deferiprone to start this spring. AISNAF pushed to expedite this trial after hearing of two NBIA families who started deferiprone treatment and saw improvements in their children.

Nardocci is planning a clinical trial for the iron chelation drug deferiprone to start this spring...deferiprone does cross the blood-brain barrier and it is hoped that it could be a viable treatment for N B I A .

Iron chelation has been tried in NBIA without much success, as previous drugs have not passed the blood-brain barrier. But deferiprone does cross the blood-brain barrier and it is hoped that it could be a viable treatment for NBIA. It has been used recently in Friedreich Ataxia with promising results. In these individuals the iron accumulates in a different part of the brain, but there is no reason to believe that deferiprone will be any less effective on iron accumulation in the basal ganglia, where it is found in NBIA individuals.

The six-month clinical study will be comprised of 10 PKAN participants from Nardocci's and Bertini's database of NBIA affected individuals, and it will have a double-blind placebo arm which means that neither the patients nor the doctors will know who gets the placebo. Pre-trial high intensity MRI's to document the iron buildup will be performed before treatment and afterward, to compare results.

AISNAF helped to get NBIA and 108 other neurodegenerative disorders included in the Italian registry of rare diseases. The medical plan for the country only covers diseases listed in the registry, and NBIA patients were not able to receive treatment and benefits previously.



AISNAF president Dr. Natalie Antonio Scalise from Rossano, Italy and vice president Lorella Faltiba from Pordenone, Italy.

The organization also organized a Scientific Workshop on Pediatric Movements Disorders in November. It was held in Rossano, Italy and sponsored by the Rotary Club there. Two of the Scientific Committee members presented papers on "The Benefits of the Baclofen Pump" and "Pain Therapy in Children."

Board president Scalise also arranged visits to various schools, city halls and other organizations in December. As a representative from AISNAF, he gave presentations on NBIA as well as educating on the difficulties of living with any rare disorder.

February 29 was Rare Disease Day in Europe. Each country had programs in place to raise awareness of rare disorders. AISNAF helped promote the various activities being held in Italy.

For more information about the organization, visit the Web site at www.aisnaf.org.

We expect to work closely with AISNAF as we have with our German affiliate, Hoffnungsbaum e.V., to help promote research and public awareness about NBIA.

Directors

(continued from page 1)



Michael Cohn from St. Louis Park, Minnesota is Director of NBIA Adult Programs.

Cohn has lived with NBIA since childhood and has a bachelor's degree in Marketing Education and a master's degree in Human Resources Development.

Cohn is also the Minnesota Branch Manager for the non-profit Hope's Hope, based in North Carolina.

Anyone who wants to participate in this program or make suggestions is asked to

e-mail him at cohnpa@juno.com or send a letter to him at 2226 Oregon Court, St. Louis Park, MN 55426.

Dianne Gray of Naples, Fla. will be director of Life Transitions and assist those who have lost a loved to NBIA. Her son Austin, 14, passed away in February 2005 because of NBIA symptoms.

She hopes to link other bereaved loved ones on the Web and create a virtual "coffee house" where people can grab a "coffee" and chat. She also plans to create a weekly blog on topics written by different participants. It will be mostly inspirational and emphasize what makes each person unique.



Dianne Gray from Naples, Florida is Director of Life Transitions Program.

Gray plans to list resources on the site, such as movies that soothe the soul, music for healing and books that are helpful. She'll include a calendar of events, such as Compassionate Friends Candlelighting Ceremony and the Bereaved Parents National Conference.

healthy while grieving or under stress.

Gray has spent the past 20 years as an entrepreneur and international business consultant. She co-founded a successful business venture in Southwest Florida and created four nonprofits that provide services for hundreds of terminally ill or disabled children. Her projects include work and travel experience in Latin American and Europe. She also served on the NBIA board from 1998 to 2004.

Gray graduated from Western Carolina University in Cullowhee, N.C., with a degree in Sports Management. She is a marketing specialist with Quality of Life Publications in Florida, which specializes in books and newsletters that help ease the way for patients and families dealing with terminal illness and loss. She also is writing a book about medical ethical decision making and her life experience, which is expected to be published in Fall, 2008.

If you would like to participate in the program she is heading up, contact her at dbgray444@aol.com.

Sandy Leap of Haymarket, Va. will be director of the Physician Registry Program. She is creating a database of doctors who have seen NBIA patients and will be sending out information on how families can participate. We hope to have a listing of physicians that will be available on our Web site starting with neurologists and neurosurgeons familiar with deep brain stimulation on NBIA individuals. After that, we will consider expanding our database to include other specialties.



Sandy Leap from Haymarket, Virginia is Director of Physician Registry Program.

Leap graduated from Texas Woman's University and has a degree in education. She worked as a teacher in Texas, but now home-schools her daughter Brittany, age 13, who has NBIA. She and her husband Rich have been active in fundraising for NBIA since their daughter was diagnosed in 2006.

For more information, contact her at rsleaper@hotmail.com.

Megan E. Thomas of Princeton, N.J., will be director of Planned Giving. Thomas is an attorney who specializes in estate planning. She graduated from Dartmouth College in Hanover, N.H., in 1979 and

A Wellness room on the Web site will offer tips for staying fit and

(see *directors* on pg. 5)

Directors

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Megan Thomas from Princeton, New Jersey is Director of Planned Giving Program.

the University of Pennsylvania Law School in Philadelphia in 1989. She serves on several volunteer boards and is the president of Preservation New Jersey, and on the board of trustees for Princeton Senior Resource Center and Gift Planning Council of New Jersey.

"After my daughter, Sonja, was diagnosed with NBIA last summer, her father and I found the NBIA Disorders Association Web site on line," Thomas said. "I am impressed by how this small

organization seeks to educate doctors and the public, raises funds for research and most especially, creates a network of family support. I have recently added NBIA Disorders Association as a beneficiary of my estate in honor of my daughter, and am encouraging my parents to consider the organization in their planned giving too."

A bequest under your will or from your revocable trust is the simplest way to make a gift of future support to the NBIA Disorders Association. You may include language similar to the following in your will or trust:

"I give (the sum of \$____ or ____ percent(____%) of my residuary estate) to NBIA DISORDERS ASSOCIATION, INC., a not-for-profit corporation located in El Cajon, California, for its general uses and purposes."

Most Americans die without a valid will. In those cases, the state governs how your property passes, but that doesn't always meet the needs of survivors, let alone meet your charitable intentions.

If you would like additional information on how to make a planned gift to NBIA DISORDERS ASSOCIATION, contact Thomas at met@stevenslee.com .

(The information contained in this article is not intended to be legal or tax planning advice. Consult your attorney or tax advisor for estate planning advice.)

If there are other services you would like to help our organization provide, you may contact Patty Wood at pwood@NBIAdisorders.org. We are always looking for volunteers!

2007 was good but expensive

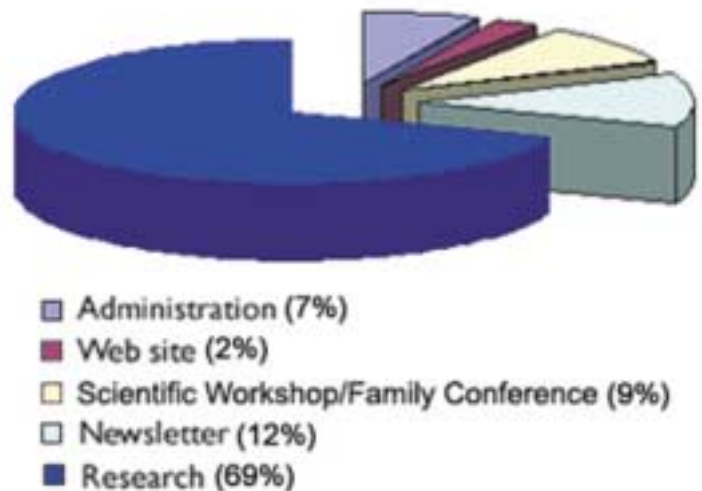
By Jeff Doerner

It was a good year: our fourth family conference was held in 2007. Three new research grants were awarded in September. But success does not come cheap.

In 2007, we took in about \$108,500 in donations, but our expenditures were about \$163,750. Even though we spent about \$43,000 more than we earned in 2007, our association is financially sound. We still hold balances in our accounts.

The pie chart shows that most of our expenses went to research, which included grants to scientists and BioBank expenses, totaling about \$113,000.

NBIA Disorders Association 2007 Expenditures



Other big-ticket items were the newsletter, which cost about \$20,000 to publish and mail three issues each year; the family conference, which cost \$15,000 and was self-supporting through sponsorships, scholarship donations, merchandise sales and registration fees; and the Web site and our online services, which cost less than \$4,000 to maintain for the year.

The association's management costs were less than \$12,000. We keep them as low as possible because all of our officers and board members are unpaid volunteers. Our largest management expenses are for an independent CPA financial review and for state registration fees, which are both required expenses when doing fundraising in various states.

(see finances on pg. 10)

Sister describes living with NBIA for herself and brother as young adults

By *Melissa Carter*



Brian Carter, 21, and Melissa Carter, 23, from Ontario, Canada. Both were diagnosed with PKAN 6 years ago.

Looking back, there were signs that my brother, Brian, and I were heading to a medical crisis. But no one thought of NBIA or PKAN when we were kids. That diagnosis wouldn't come until much later.

I remember when we were children growing up in Blackstock, Ontario, where we still live in Canada. Brian fell a lot. He also stuttered and went to speech therapy. As soon as he would stop the speech therapy, the stutter would come back.

I didn't have any problems speaking — not then. But I had trouble swallowing and went to therapy for that.

The real trouble began six years ago, when Brian was 15 and I was 17. His gait changed. He started dragging one foot, prompting visits to many doctors and numerous tests. Finally, at Sick Children's Hospital in Toronto, doctors said his symptoms meant he either had Wilson's disease, which they could manage with drugs, or the

incurable Hallervorden-Spatz Syndrome, the former name for NBIA and PKAN. Doctors sent his blood to Chicago for a DNA test because researchers in Oregon had recently discovered the misspelled gene for PKAN. The results came back: Brian had Hallervorden-Spatz.

I remember getting the news: My mom and I both started crying because we couldn't believe it. Brian was going to need a lot of help. At the same time, I was having problems with my hands and my jaw. I thought maybe it had something to do with getting my wisdom teeth pulled and wearing a retainer. Doctors ordered the same DNA test of my blood. Their suspicions were right. I had PKAN too.

My parents were tested, and each of them had the misspelled gene. Our older sister was found to be a carrier, but she does not have the disease.

The diagnosis was like a nightmare that had become real. The disease wasn't going to go away without a miracle. Brian and I were only the second cases doctors had ever seen at Sick Children's; the first was a long time before us.

But good things came to me and Brian, too. While at Sick Children's, we each received a wish from Starlight Wish Foundation. I chose a shopping spree at the Eaton's Centre and got to stay in a very nice hotel in Toronto. A limousine toured me around the city. Brian chose a trip to Disney World in Florida. We each had an amazing time.



Brian and Melissa with their parents, Don and Carrol Carter at home in Blackstock, Ontario Canada.

We then went to Bloorview Kids Rehab, a center for disabled children, for therapy. We met wonderful doctors, nurses and

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Carter

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therapists. When I was in rehab I did exercises to strengthen my legs and I still do them. They help me walk on my feet flat because sometimes I walk on my toes.

Brian went to therapy for his walking and did exercises to help him walk straight and strengthen his legs and back. He went into speech therapy again where he learned how to talk slowly and clearly.

Most of our therapy lasted for about a year to a year and a half and was once to twice a week, although Brian's leg therapy initially was three to four times a week. It was very beneficial. He went from leaning back far when he walked to walking almost normally.

My swallowing therapy worked very well. I have no problems swallowing, although I have trouble chewing because of weak jaw muscles.

We also got great care at Toronto Western Hospital and went to Oregon for a study and a battery of tests, including eye tests, blood tests, MRI's and lots of psychological tests. We had tons of forms to fill out.

In Oregon, we met Dr. Susan Hayflick, Dr. Penny Hogarth and Allison Gregory. We also met some of the researchers who work behind the scenes on our disease, which was nice for us and them. They could put faces to the research. The food was great, and Oregon was beautiful. From the hospital, we had a view of the mountains, the ocean and huge trees.

I had been getting Botox injections in my jaw but stopped because they were very painful and not much help. I also never had a speech problem until I was a university student and the Botox got into my voice box and temporarily paralyzed part of it. I was sitting in class and went to answer a question and could not say the letters B, S and then about half of the alphabet. After the Botox wore off, I still had trouble speaking and I asked my teachers not to ask me questions in class, and they didn't. I still stutter sometimes, and when I need to repeat something, I can have a hard time getting it out, or my voice can be very soft.

Dr. Hogarth suggested I get Botox in my neck muscles, and those injections worked, although they paralyzed the front of my neck and I had problems lifting my head. Dr. Hogarth also suggested Artane, so I quit the Botox and am still on Artane, as well as Baclofen and

B₅ vitamin. I am doing quite well.

Brian is also on Artane, Baclofen, B₅ and Clonazepam and is doing great. Brian also gets Botox in his leg and foot and finds it really helps him walk and relieves pain from muscle tightening.

Brian considered having deep brain stimulation surgery but is going to wait and see if his symptoms worsen. I was told that I would not qualify because my symptoms are not bad enough but I may want to reconsider if or when I get worse. I seriously don't think I could do it unless my symptoms were extremely bad.

Brian is now 21, finished with high school. I am 23 and have a degree in medical administration from Durham College in Oshawa, Ontario.

"We attended the NBIA Family Conference in May 2007 in Cincinnati and had a blast. I now e-mail people I met from all over the world – from India to Iceland to Texas – and as close as three hours from home."

Right now, I am doing child care and volunteering at a local hospital in the day surgery admissions, and handing out water. Brian and I are both working with an organization that helps disabled individuals find and keep jobs. I have an interview with a doctor, so I am excited about that. Brian is expecting to find work soon as well.

My favorite things are reading, shopping, hanging out with friends, going on the Internet, chatting on the phone, watching TV and movies. Brian enjoys playing video games, watching TV, hanging out with friends and the Internet.

One of the best things that we have done is connect with the NBIA Disorders Association. We read the newsletter, and my Mom gets e-mail updates.

We attended the NBIA Family Conference in May 2007 in Cincinnati and had a blast. We saw doctors and researchers from Oregon again and met families who have the same problems we have. We learned about therapies that have helped others and made new friends. I now e-mail with people I met from all over the world — from India to Iceland to Texas — and as close as three hours from home.

Through all of this, the only way my family and I have been able to deal with PKAN is through God and prayer. Everyday we pray for everyone who is suffering along with us. We believe that one day there will be no such thing as PKAN and NBIA, that it will be treated with a simple pill and that there will be no symptoms and no more pain for any of us.

Tasty fundraiser brings in nearly \$1,600 for research

By Kristi Ose

My husband and I found a tasty way to benefit the NBIA Disorders Association Research Fund. We hosted a wine tasting at Stoney River Restaurant in Louisville, along with a silent auction, and raised \$1,595.



Kristi & Ron Ose of Goshen, Kentucky at wine tasting fundraiser for NBIA Disorders Association Research Fund.

Sixty people paid \$25 for the event, which included a \$10 voucher toward dinner. Part of the dinner proceeds and \$5 from the admission went to the association. Each month, the restaurant assists a charity in this way.

Guests had four wines to choose from and a nice selection of hors d'oeuvres. I spoke about the NBIA Disorders Association and the progress that is being made with research. Brochures were placed on every table to spread awareness.

The silent auction portion, which consisted of 12 items, brought in more than \$500. Others who couldn't attend gave direct donations. It was a fun night and worth checking out restaurants in your area to see if they would be willing to put on this type of event.

Silent Auction Donors

Patti Allen
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Betty Grant
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Todd & Chris Richards
Stoney River Restaurant
Salvatore & Alicia Ruriani
Tim & Laura Snyder



Guests at the wine tasting fundraiser in Louisville, Kentucky. From left: Sue Marshall, Phyllis Ose, Rita Pipes, Betty Grant, Serena Middleton and June Creek.

May NBIA fundraiser in Virginia will kick off "Ride to the Wall"

Motorcycle enthusiasts from across the country who ride to the Vietnam Veterans Memorial in Washington, D.C., May 25 can make a pit stop in Virginia to help the NBIA Disorders Association.

The Haymarket Masonic Lodge is hosting a breakfast from 6:30 to 8:30 a.m. the morning of the "Ride to the Wall," which is being held to honor veterans who lost their lives in the Vietnam War. The lodge is at 6711 Jefferson St, Haymarket, Va. (20168), and the breakfast is free to all motorcyclists.

Donations will be accepted on behalf of NBIA Disorders Association, and the lodge will match contributions up to \$5,000. Members of the lodge are friends of the family of Rich and Sandy Leap, who have a daughter with NBIA.

For further information about the fundraiser, contact Bruce Silvernale at 703 489-6408. For more information about the lodge, see the Web site at www.haymarket313.org.



"You are cordially invited to participate in a Pampered Chef national fundraising event to support NBIA Disorders Association Research Fund."

Here's a unique NBIA fundraiser you can take part in from home

Calling all chefs.

As a way to raise money and awareness for the NBIA Disorders Association, shop for yourself or buy that special cook in your life a high-quality kitchen tool or some other gift from The Pampered Chef. All proceeds for this national fundraiser will benefit the NBIA Disorders Association.

During April and May, our NBIA community and everyone they know can buy kitchen tools as well as buy gifts for every occasion from The Pampered Chef by going to a special online site. NBIA parent Sandra Leap of Haymarket, Va. is chairing the effort in cooperation with Pampered Chef consultant Doina Heinz who is donating all of her time and profit to this event.

The two women enjoyed great success in September 2006 when they worked together on a cooking show fundraiser in Northern Virginia. With contributions from just 15 participants, the show yielded almost \$1,200 in sales. Although The Pampered Chef corporate fundraising program offers a profit of 10 percent to 15 percent, the consultant donated additional funds to create a 20 percent return of almost \$250.

Seeing the potential, Leap and Heinz are launching this national effort. This time, Heinz is working at a non-profit level, so we expect close to a 40 percent return on every dollar spent.

The money will support the association's Research Fund.

Visit www.pamperedchef.biz/doinaheinz and select Our Products (bottom left), and under organization, enter NBIA. For questions about products and help ordering, contact Heinz at DoinaHeinz@comcast.net.

Spread the word by sharing this information with your entire contact list. We hope everyone will join in.



Pampered Chef deep dish pie plate



Pampered Chef mini baker

Mark your calendar and join us for Sept. 22 Leap golf marathon

By Rich Leap

It's time again for the third Queen Bee Golf Marathon. The 2008 event is set for Sept. 22 at Piedmont Country Club in Haymarket, Va.



Last year you helped us raise the bar tremendously. The day was a great success. We invite you to join us for another fun filled day of golf and fellowship. For information on golfing, sponsorship opportunities, donation of prizes or supporting our golfers, log onto www.queenbeeworld.com or e-mail Rich or Sandy Leap at rsleaper@hotmail.com.

Won't you BEE THE ONE to help, to share, to care to find a cure?

NBIA merchandise or our cookbook make perfect holiday gifts for the teachers, aides, nurses, therapists, family and friends on your list. To order visit our Web site at www.NBIAdisorders.org



Finances

(continued from page 5)



*Jeff Doerner, Treasurer
of NBIA Disorders Association*

At year-end, our General Fund's bank account held about \$32,500, so we are able to meet our operating expenses. The year-end balance in the Research Fund was about \$124,000. That large amount is to make our research grant payments of \$70,000 in 2007, plus meet BioBank expenses of about \$20,000 per year. That leaves

enough for another research grant, so we have published a Request For Proposals to the scientific community for 2008. Any fundraisers for the Research Fund held this year could also increase this total so that additional grants may be available.

Our 2007 financial statement and tax return is published on our Web site. I invite you to review the documents for more in-depth information.

In 2007, the average contribution was \$169 and half of all donors gave \$50 or less. Our largest single donation was \$5,000, and there were 20 donors of \$1,000 or more. Our smallest donation was \$2 and that is very special to me even though I do not know the donor. To me it means that no matter how large or small, every contribution counts and helps us move closer to our goals.

Requests for Proposals for grants for clinical research due June 13

The NBIA Disorders Association is accepting applications for one-year grants for clinical research studies related to the early detection, diagnosis or treatment of patients with NBIA.

The application and instructions can be found at our Web site at www.NBIAdisorders.org under the Grant Info link on our home page. The deadline for letters of intent is June 13. The grant(s) will be awarded in November.

The purpose of these grants is to encourage meritorious clinical studies designed to improve the diagnosis or therapy of Neurodegeneration with Brain Iron Accumulation. The research will be conducted in the United States, Europe, Canada, Australia, New Zealand, Brazil, Argentina, Chile, South Africa, Japan or Israel, and where supervision of grant administration is possible.

Grants will be awarded to academic researchers to initiate small clinical trials, the results of which could be used to obtain funding from the National Institutes of Health, Food and Drug Administration or other funding agencies, or to attract a corporate sponsor.

Procedures or proposed therapeutic trials may be new, based on recent biochemical or pharmacological evidence, or be in preliminary states of clinical investigation.

For more information, contact Patricia Wood at pwood@NBIAdisorders.org.

Fourth NBIA Family Conference in Germany scheduled for the end of May

By Angelika Klucken

The Fourth NBIA Family Conference in Germany sponsored by Hoffnungsbaum e.V. is set for May 30 to June 1 in the little village of Horath, near Trier, at the Family Hotel Hochwald where the last three were held.

The conference motto will be "Patients Contribute to Research." Neurologists from university hospitals in Cologne, Göttingen, Munich and Innsbruck (Austria) will be on hand to launch their projects.

(see conference on pg. 11)

Hoffnungsbaum's funding pays off: Klüh-Prize for Timmermann project

By Angelika Klucken



*Dr. Lars Timmermann receives research grant from the Klüh Foundation in Germany.
From left: Dorothee Timmermann, Lars Timmermann, Eva Luise Köhler,
the wife of Germany's Federal President and donor Joseph Klüh.*

Dr. Lars Timmermann's research into deep brain stimulation in NBIA patients is taking another leap forward.

After receiving the first-ever research grant worth 12,000 euros last year from the NBIA Disorders Association's sister organization in Germany, Hoffnungsbaum e.V., Timmermann later won a foundation research grant to take his work to the next level.

The first grant, to Timmermann and his staff at the University Hospital of Cologne, was to systematically collect and compare data on deep brain stimulation done on NBIA patients from around the world.

The data looks at timing, settings, adjustments, results, side-effects, complications and other issues that will help families make decisions about whether to seek DBS for their loved one. Results of that retrospective study are almost completed.

The second grant will enable Timmermann to follow patients in real time, going forward to see how they respond to DBS. That grant is from the Klüh Foundation,

which has been making annual grants for "Innovation in Research and Science" since the mid-1980s.

The foundation's grant to Timmermann, made late last year, was for rare disease research, in cooperation with Germany's recently launched National Alliance for Chronic Rare Diseases. More than 50 applicants competed for the prize. Eva Luise Köhler, the wife of Germany's Federal President, Prof. Horst Köhler, awarded the Klüh-prize to Timmermann in the presence of the donor, Joseph Klüh.

Conference

(continued from page 10)

Patients with PKAN can contribute through blood draws intended for a study that will investigate a diagnostic tool for another rare disease called Neuroacanthocytosis (NA) and a possible connection between PKAN and NA. Some families will also be filling out medical history forms for the NBIA Disorders Association BioBank and will take part in a neuropsychologist's poll examining cognitive aspects of the disorder. Another of the conference goals is to find out whether sonograms could be a reliable diagnostic tool for NBIA. Additionally the possible value of metabolic profiling with a tool called "metabolomics" will be introduced to the audience.

Families will have opportunities to get together, exchange experiences and make friends. Volunteers will care for the children once again. Arrival is on Friday afternoon, the main conference is on Saturday, and the association will have a general meeting Sunday morning.

NBIA families in Europe who want to attend may contact Hoffnungsbaum e.V. by e-mail at hoffnungsbaum@aol.com or by phone at +49/2051/68075 for information and registration. Documents are in German. Correspondence or calls in English are possible to clarify questions. Language at the conference will be in German, but foreign guests are invited to come with their own interpreters.



Melissa Ann Moore

F ebruary 7, 1982 - D ecember 17, 2007

*Gone from our sight, but never from our memories,
Gone from our touch, but never our hearts...*

In Loving Memory

*A my Miller
March 29, 1985 - J anuary 22, 2008*

*Mandy Miller
March 13, 1982 - F ebruary 4, 2007*

*Susie Miller
October 12, 1986 - D ecember 2, 2002*

It is so hard to believe our three girls are gone. Their suffering is over with and we know they are safe in the arms of Jesus.

We will never forget their sweet smiles, their charming personalities and their cute senses of humor. We feel blessed to have had them for all these years. How we miss them.

David & Mary Miller
Anna & Andy Byler

Truly the Children of God: A tribute to the Miller girls

By Dianne Brown

It is with great sadness that I write this memorial to "my three Miller girls." Amy passed away on Jan 22. Her sister Mandy died last year, and Susie passed away in 2002. I was their teacher for most of their school years, and they will forever live in the hearts of all they touched.

"Prepare to fall in love." That was a comment told to me when Mandy Miller was transferred to my pre-school classroom. Never did I expect the rewards of what was to come. I had before me the most beautiful child. She had a warm and giving smile that captured your heart. No one knew at that time why she was stumbling often and why her two younger sisters were having the same difficulty.

We were triply blessed when Amy and Susie enrolled. But soon after that, they were diagnosed with NBIA, then called Hallervorden-Spatz. Everyone at the special education school was devastated and we held counseling sessions with hospice and trained counselors. We all shed countless tears.

Their condition required unique techniques and goals with constant revisions for their ever-changing positioning and comfort. They lost their mobility, verbal communication and ability to help themselves, but their mental abilities were intact and continued to accelerate.

Adaptive communication methods were employed and the girls never lost their ability to relay their feelings to us. Trachs, G-tubes and implants followed, along with nursing staff. But each day of their lives they seemed to give more to us than we gave to them.

To love and make every day fun and full of sunshine became the motto of my classroom and we all became better people because of these girls. Because of the special needs, the girls stayed in my classroom for most of their educational careers. Yet, they were not secluded. They were extremely social and were known and loved by the whole district. I truly believe that staying in school extended their lives.

Each girl was unique and caring. Mandy, the oldest, endured all kinds of treatment and trial recovery methods. She suffered more than the others and we all cried so many tears for her as she suffered. Amy was the soft, motherly child who loved her baby dolls and was often given babysitting responsibilities for other children in the class. She took the duty seriously. Susie was the feisty one with an extremely strong personality who loved to play jokes. She seemed to fight her illness the hardest, which often caused greater discomfort. But she hated what was happening to her.

All died peacefully. Susie with Amy holding her hand; Mandy, gracefully and thankful to others. Amy was in a coma for a short time before

(see tribute on pg. 13)

Tribute

(continued from page 12)

her death but awoke with a smile on her face to tell us a last goodbye.

My heartfelt sympathies to Mom and Dad Miller who gave of their lives for so many years to care for the girls. They are such beautiful people who never questioned their purpose. My love to Anna, their younger sister who is expecting her first child in June. She was a major caregiver with always a smile on her face and never a complaint.

God allowed us to hold and love these girls for a short time but the impact they had on our lives will last forever. They enriched us all.

My Wishes

Reproduced with permission from *Aging with Dignity*

It is never easy when someone we love becomes seriously ill, and it is even more difficult when that person is a young child. There is understandably an emphasis on getting the most accurate diagnosis and obtaining the best medical care possible. Even if the child is not yet old enough to make medical decisions, it is important to talk to the child about what they want, what makes them feel safe and comfortable, and what they want their parents, doctors, and nurses to know. These can be difficult topics to broach.

A new document called *My Wishes* makes these difficult conversations easier. Based on the popular *Five Wishes* advance directive, which is used by more than 11 million adults to express their healthcare preferences, *My Wishes* helps identify what is important to children who are facing a serious illness. *My Wishes* and *Five Wishes* are both offered by Aging with Dignity, a national nonprofit organization with a mission to safeguard the human dignity of those who are most vulnerable, especially those who are seriously ill.

"Being the parent of young children myself, I can understand the sensitive nature of these conversations," says Paul Malley, president of Aging with Dignity. "*My Wishes* puts the discussion in terms that are meaningful and understandable for the children — and it provides a guide for parents, caregivers, and health providers to address these important issues without starting from scratch."

My Wishes is a simple booklet developed by child life professionals that helps children express their wishes in their own words. Laid out in a colorful document with plenty of space on each page for children to draw or write stories, *My Wishes* includes the following examples:

Wish #1: My Wish for how I want people to treat me

"I wish to be cared for with kindness and cheerfulness; I wish to



My Wishes document provided by Aging with Dignity, a non-profit whose mission is to safeguard the human dignity of those who are seriously ill.

have my hand held and to be talked to whenever possible." Children are given space to affirm these and other statements about the type of compassionate care they would like to receive. They are given the opportunity to draw pictures of their favorite things that they may want to have with them.

Wish #2: My Wish for how comfortable I want to be

"I do not want to be in pain; These things make me feel good; These are some things that I do not like." Wish #2 is an important tool in helping understand children's approach to pain management. Many people have different approaches to pain management, especially when they may become tired as a result.

Wish #3: My Wish for what I want my loved ones to know

"I wish to have my family and friends know that I love them; I wish for all of my family members to be nice to each other." *My Wishes* deals with more than medical issues—it helps to address important emotional issues, including love and forgiveness.

Wish #4: What I want my doctors and nurses to know

"I want you to call me by my name or nickname; I know I need time to rest and sleep, but I also want to have time to play." Wish #4 helps children explain to their health care provider how they want to be addressed, how they want to be talked to about medical issues, and assert that they would like measures taken to respect their dignity and privacy.

My Wishes speaks to children in language that is easy to understand, saying that "there are many things in life that are out of your hands.

(see *wishes* on pg. 15)

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

Julie Ardery
Mary Ann Roser

Belcher Family
Jayne Prendergast

Eric Brolin
Bill & Laura Brolin

April & Tracie Flinn
Bud & Althea Brown
Joyce Judd
June McClure

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Nadine McDade
Renee McDade
Samantha McDade
Gary McHenry

Drs. Baker, Gardner & Hunter
Bill & Kim Patterson

Brent Bonfiglio & Family
Angela Bonfiglio Allen
Norma & Patricio Dauguet
Angela Dragotta
Theresa Foster
Allison Mochizuki
David & Louisa Mochizuki

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Mary Tapke
Saroja & Chandra Ullagaddi
Mary Walsh

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Diana Deene
Nancy Overman
Sara Phillips

Cynthia Melton
Christian Clem

Jared Ose
Tim & Laura Snyder

Keri Patton
Paul & Janet Buhay
Cyndy Patton
Charles & Florence Waniak

In Honor Of

Brandon Richards
Richard & Georgette Peckham

Mary Ann Steinker
Angela Steinker

Kimberly Wood
Gary & Barb Hampton
Carole McDowell
Dr. Rick Rader
Robert & Jean Starling

Mary Ann Roser
Audrey Lee

Jonathan Stretter & Family
James & Ellen Dick
Diane & Wally Ingram
James & Sandra LeVos
Ron & Donna Stetter

In Memory Of

Alyssa & Sabrina Barbiero & Family
Sciocchetti Family
Pete & Steph Sherman
Scott & Janice Smith

Clay C. Crews
Julia Richards
Mark & Martha Nell Richards

Joyce Knotts
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Hendrik & Delinda Luinge
Patricia Moore
Betty Twiss

Wade Patton
Paul & Janet Buhay
Cyndy Patton

Julie Smyth
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Nancy Greyer
Carol Lindeman
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Bruce Belcher
Chris Cazzato
John & Dorothy Magee

Sharon Denise Haugen
Anonymous
Kathy Kelchner

Ralph Hawkins
Pat Jordan
Harry & Verna Krebs

Edward M. Malcolm
Mark & Martha Nell Richards

Mandy, Amy & Susie Miller
David & Mary Miller
Brenda Wolcott

Ben Patterson
Charlotte Kittler
Bill & Kim Patterson
Ray & Linda Patterson
Geneva Pounders
Sloan & Daniel Powell
Glinda Richardson
Clara Jane Slaughter

Richie Roberts
Bernie & Judy Roberts

Frank P. Venuto
Brad Venuto

MESSAGE FROM THE PRESIDENT



Patty Wood

I am very excited to say that this issue of the newsletter is the first that brings you news from both of our sister organizations in two other countries, Germany and Italy. These non-profits have been working hard to educate the public in their countries about NBIA and to promote research into the disease.

While we are three separate entities, each with our own legal status and country laws to follow, we are united in our cause — a cure for this devastating disease. We know that by working together much more can be accomplished than by going it alone.

In 1996 when I founded what is now the NBIA Disorders Association, it was because there was nowhere for families to turn for support and up-to-date information on NBIA (then called Hallervorden-Spatz). There was no organization in the world supporting research into our disease, and few researchers were interested.

Now, just 12 years later, there are three very active, involved organizations working together, and more researchers than ever are interested in studying NBIA. We have made many inroads into finding answers and are looking at different therapies that did not exist when we started. But there is still much work to be done.

I encourage NBIA families from other countries to seriously consider talking with other families about starting a non-profit in their country to work with us to continue this expansion of awareness and support internationally. Because of language and distance barriers, there are still families that we cannot adequately support. Also, it's much easier for someone living within a country to promote public awareness than it is for outsiders to do it.

NBIA is such a rare disease it can be difficult to get adequate funding to promote large programs. One solution is to create many smaller groups in each individual country all working together to create a larger network of information, outreach and research opportunities.

The German group, Hoffnungsbaum e.V. has made significant contributions to research studies, including providing \$10,000 to the NBIA Disorders Association's Research Fund to go towards a grant. They also funded a grant themselves this past year to gather information on Deep Brain Stimulation for NBIA individuals. The more groups that are supporting the efforts of researchers, the better the odds of discovering new therapies and possibly, a cure.

We can be proud of the achievements of our three organizations, and we hope that more will join us in the near future. What a difference you could make!

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.

Jeff Berwick	Charles & Karen O'Neil
Slava & Maria Buchkovich	Jerry Oshiyama
Christine Buhay	Sally Phillips
Lillian Cady	Diane Raymond
Don & Marie Cullen	John Charles Rush
Paul Deltry	Responsive Solutions Inc.
Cheryl Dendquist	Linda Rocklin
George Fabe	Sacred Heart Church
Stephanie Freda	John Schenck, MD
John Gerland	Debora Stalker
Teresa Ghianni	Carol & Gary Steinker
Haire-Bohmer Mgmt Group	Christopher Stretter
Jim Hayes	Mary & Tom Tapke
Fred & Betty Knodle	Megan Thomas
Sebastiao Martins	Paul Tierney
Lori Mitchell	Rose & Rod Walker

Wishes

(continued from page 13)

My Wishes gives you a way to control something very important — how you are treated if you are very sick. It is easy to complete, and helps you say exactly what you want."

Amy L. Blais, Pediatric Hospitalist at Central DuPage Hospital in Winfield, IL, offers an example of how *My Wishes* has helped children, parents, and health care providers communicate:

"One night in the PICU the condition of one of our pediatric patients became acutely worse. A copy of *My Wishes* was available on the unit. I spoke with the parents outside of our patient's room and then brought the booklet into the room. As a group, the patient, his parents and I worked through the issues. Having a chronic, potentially fatal illness, my patient had thought about many of the issues before and had strong opinions as to what he wanted.

With the help of the *My Wishes* booklet, I was able to answer his questions and address his concerns. Prior to that night, he was not aware that they could become part of the medical record and be heeded by the medical team. It was a comfort to his family and the staff involved that his wishes were followed."

My Wishes can be given to a child by a parent, a caregiver, or a healthcare provider. The time to introduce *My Wishes* can be after a new diagnosis or a change in condition, or as a preparation for an upcoming hospital procedure. Any time when important medical decisions are being made in the life of a seriously ill child, *My Wishes* can help make these conversations a little easier.

To get copies of *My Wishes*, visit www.agingwithdignity.org or call (888) 594-7437.



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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

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Visit our Web site at

www.NBIAdisorders.org

from discovery to cure

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