

Hayflick lab faces closure

By Patricia Wood

Dr. Susan Hayflick — NBIA's top researcher for the past 17 years — said she faces the prospect of closing her lab by the end of this year after being turned down twice in two years for funding by the National Institutes of Health.

Hayflick said she hopes to avert a closure if funds can be found elsewhere, but she realizes the amount is daunting: \$250,000 by the end of the year. She, along with the NBIA Disorders Association, is asking NBIA families, politicians and anyone else who will listen to help.

Hayflick learned in late June that her latest application for an NIH research challenge grant at the Oregon Health & Science University was rejected, and she is fearful her second will suffer the same fate later this month.

In March NIH offered approximately \$400 million in challenge grants to researchers as part of an additional \$10.4 billion designated to NIH with stimulus funds, and were flooded with 18,000 applications. Only 2 percent will be awarded grants.



Dr. Susan Hayflick from Oregon Health & Science University discusses research funding issues at family conference.

Hayflick said she hopes to avert a closure if funds can be found elsewhere, but she realizes the amount is daunting: \$250,000 by the end of the year.

The rejection comes as NIH did not renew Hayflick's regular grant award for studying NBIA this year. She has applied twice to renew this grant. That leaves her lab without its main source of support. Even if Hayflick applies for a new grant in the next NIH cycle in October, it typically takes two years for the money to flow in. By that time, her lab researchers will have moved on, making it all the more difficult to try and rebuild, she said. *(see Hayflick on pg. 3)*

Fifth International Family Conference biggest ever, praised by participants

By Luann Rein

Our Fifth International Family Conference in downtown Indianapolis drew 115 participants from eight countries, making May's biannual gathering the largest and one of the most successful ever.

Seventeen new families joined veteran conference attendees representing the United States, Canada, The Netherlands, Germany, England, Italy, Iceland and India. *(see conference 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.

Hayflick

(continued from page 1)

After seeing big increases in research funding between 1999 and 2003, when the budget rose from \$15 billion to \$26.4 billion, NIH funding has been flat in recent years — \$28 billion in 2004 and \$29.5 billion in 2008. President Barack Obama's 2010 budget request is \$31 billion, a 1.4 percent increase over 2009. Sustainable and predictable funding increases for biomedical research are crucial to progress in fighting diseases, but NIH budgets have failed to keep pace with the cost of inflation and conducting biomedical research according to an index used by the government to measure the rising cost of equipment and research workers.

While many researchers are feeling the pressure of lost grants and no doubt some have closed their labs, the loss is much more devastating for the rare disease research community.

As Hayflick put it in a recent letter to the Office of Rare Diseases Research at NIH:

"I write at this time because of the crisis facing the rare disorders community. With NIH extramural funding rates at an all-time low, biomedical research labs are closing. These losses will have little measurable impact on investigator communities in cancer, hypertension, diabetes, etc. But for many rare disorders, the entire research community comprises only one or two investigators. If they are lost, research comes to a halt. Moreover, the investment needed to rebuild from the ground up will be enormous. This is an especially demoralizing time for investigators, and the impact of lost productivity will be hard to quantify.

This crisis is personal for me and will impact members of the NBIA Disorders Association, our family advocacy group. I have been continuously funded by the NIH for 14 years, focusing my studies on this group of rare disorders. My NIH funding has ended and I have little hope for renewed funding in time to preserve the expertise in my lab group. The fallout on the NBIA research community of my lab's closure will be deep and long-lasting. I want to be sure that our friends, colleagues and advocates at ORDR recognize and acknowledge this crisis. We need your help."

Fewer than five researchers devote a substantial amount of their careers to NBIA, so the loss of Hayflick's lab represents a significant loss for the NBIA research community. Her lab was primarily responsible for finding the first NBIA gene in 2001 and a second in 2006. She directs the NBIA Disorders Association's Scientific & Medical Advisory Board and is considered the leading expert on NBIA in the research community.

The NBIA Disorders Association is mounting a media relations campaign to help make the public aware of the dire funding situation and avert the imminent closure of the Hayflick lab. The goal is to raise \$250,000, which would keep the lab open for two years, at which time it is hoped that funding from the NIH would resume.

The amount is the minimum needed and does not include a salary for Hayflick. The NBIA board still intends to award seed grants through competitive applications in December like it normally would with funds dedicated to our Research Fund.

To raise money for Hayflick, the Association hopes to attract support through publicity on TV shows, newspaper articles and online promotions. We will have a link on our Web site home page at www.NBIAdisorders.org for donations to the "Hayflick Lab Campaign" or you can mail a check addressed to NBIA Disorders Association and put "Hayflick Lab Campaign" on the memo line or on the donation envelope included with this newsletter so we are sure to credit it correctly.

Association Executive Director Luann Rein is coordinating a Facebook cause page where donations will be accepted specifically for this cause. We are hoping NBIA families will start Firstgiving campaigns with their families and friends, and Rein is available to help them with questions on how to set up and use this medium.

If any of you know a media or public relations contact who can help us promote our cause, please contact me immediately at pwood@NBIAdisorders.org or phone me at (619) 588-2315. If you know of any foundations or philanthropic families that might contribute, I would appreciate hearing from you.

Susan Hayflick has been more than a researcher to our families over the past 17 years. She has been a friend, a tireless advocate and our brightest hope for effective treatments and ultimately, a cure. Losing her lab would be an enormous blow to our community, one that would take years to recover from. This is our chance to help her, ourselves and our loved ones.

As we demonstrated in our closing ceremony video at the Fifth International Family Conference in May, "We believe in miracles." Help us make one happen.



Drs. Paul Kotzbauer and Dr. Susan Hayflick answer research questions during a session at the family conference.

Conference

(continued from page 1)

Judging from the evaluations submitted afterward, participants enjoyed the mix of scientific topics and caregiver issues sandwiched between social events and networking time. The downtown location, amid shops, restaurants and parks was ideal for families and strolling.

Twenty-four attendees said the conference exceeded their expectations and another 12 said it somewhat exceeded them. No one said, "Not at all."

Some participants wanted more time in some sessions, such as with scientists who updated the group on their research. Others found certain information to be too US-oriented for international families. We received suggestions on how to better organize social events so families could meet earlier, and we will take all ideas into account for next time.

Once again, families had a chance to visit with scientists and doctors for medical appointments on Thursday.

On Friday morning, president Patty Wood kicked off the conference by sharing milestones since she and Dr. Susan Hayflick collaborated to found the association in 1996. The first newsletter published in 1998 had a mailing list of 75. Today, it tops 3,700!

Keynote speaker Brian Denger with Parent Project Muscular Dystrophy urged us to be advocates and gave tips for lobbying Congress. Parent Ron Stretter led a session to help us get to know each other better, and our program directors updated us on their work.

Families had a chance to share information during the traditional information exchange session, while another session focused on the need for families to support scientific research at a crucial time when federal dollars are stagnating.



Samantha Jennings from Ottertail, Minnesota enjoys the music in the care room.

Sandra Grether, a speech-language pathologist, discussed options for patients with communication needs, and participants were invited to visit a separate room with vendor exhibits, demonstrations and assistive technologies.

On Saturday, five leading NBIA researchers updated us on their work. Hayflick not only talked passionately

about her research but again stressed the importance of family fundraising and finding alternative sources of support.

Participants received details about an iron chelation trial from nurse practitioner Nancy Sweeters, who spoke about the use of deferiprone, the first iron chelation drug to cross the blood-brain barrier.



Patty Wood with Shane Murdock collecting his silent auction basket at the picnic.



Zion Coates from Birmingham, Alabama at the picnic.

The rest of Saturday was dedicated to social events. Despite the threat of rain, a picnic replete with games was held that would have been impossible without Patty's dedicated sisters and other family members, plus help from sister Diane Murdock's friends — Carol Keller, Lynn Hurlbut, Peggy O'Connor Campbell and Grova and Judy Lewis. A touching balloon ceremony to memorialize loved ones capped off the afternoon.

A dessert social that evening was highlighted by a silent auction for the remaining gift baskets that had not be raffled off or auctioned at the picnic. Most families brought baskets reflecting their state or country, and all of the money raised went to the association.

On Sunday Dr. Ajay Perumbeti spoke about gene therapy, and I was introduced as the organization's new executive director.

During the conference, those in our care room were entertained by pet therapy dogs and musical therapists. Volunteers Lindsay McDaniel, Hayley Marie Rabbers and Nadine, Samantha, and Amy McDade assisted throughout.

The closing ceremony, "We Believe in Miracles," was a moving tribute of photographs and music. Bill Hartwell and I rewrote the words and changed some of the rhythms to the song, "I Believe in Miracles," and board member Veronica Bonfiglio assembled a montage of quotes and pictures. Board member Sue Laupola and friend Tom Ford took photos and Tom assembled the package into an unforgettable closing.

Keynote speaker urges: Let your inner advocate come out

There is a little bit of the advocate in all of us, Brian Denger told parents and others at the opening of the Fifth International NBIA Disorders Association Family Conference in Indianapolis in May.

The advocate inside of Denger came out when the school where he wanted to send his son in Maine denied young Matthew access, saying it "couldn't meet the needs" of a child with Duchenne muscular dystrophy, Denger said during his keynote speech. He promptly filed a discrimination complaint. And won. "One little boy made quite an impact," Denger said.



Brian Denger, from Biddeford, Maine, with his two sons who have Duchenne muscular dystrophy. He was the keynote speaker at the family conference.

So did Denger. He became the parent advocate coordinator for Parent Project Muscular Dystrophy to help others. Today, he is a regular on Capitol Hill, button-holing members of Congress.

Parents can be excellent

advocates, he said. They often have heart-rending stories to tell, and members of Congress and state legislatures are often touched by what they hear. Many lawmakers are parents, too.

Denger encouraged parents of NBIA children to consider lobbying and offered the following tips:

- *Go with a strategy and a plan. He has a daughter with diabetes, and when he learned one of his representatives is a supporter of diabetes research, he thanked her for her work in supporting diabetes research as a way to find common ground. Building relationships is important, he said.
- *Be nice. "We do get more flies with honey than with

vinegar," Denger said.

- *Be concise. You probably will get 20 minutes. Stay on message. Most of the time you will get a staffer in their 20's.
- *Leave a picture of your child.
- *Dress appropriately, not in jeans and sweat pants.
- *There is nothing wrong with tears and emotion.
- *Always remember to say, "Thank you" and send a "thank you" note.
- *Always get contact information for follow-up purposes. Stay on their radar. Sooner or later you will develop a champion.
- *Partner with other organizations. "There is value in numbers," Denger said.

It can be intimidating, he said, but it can also be rewarding. In the latest federal budget, he and others succeeded in securing \$200 million for muscular dystrophy research, he said.

Denger's advice was helpful to the Leap family who had appointments with the Oregon and Virginia senators offices in Washington D.C. on July 16. Their purpose was to make these representatives aware of the NBIA community and how the loss of rare disease researchers such as Dr. Susan Hayflick would be so detrimental. The Oregon senators were very helpful and sent a letter to the Senate Appropriations Committee supporting increased funding for rare diseases and specifically NBIA.

Board withdraws from Genetic Alliance BioBank; sends samples to Oregon lab

Faced with escalating costs, the NBIA Disorders Association board voted in April to withdraw from the Genetic Alliance BioBank.

The association was a charter member of the bank five years ago, but its Scientific & Medical Advisory Board endorsed the idea to withdraw. Both boards agreed the samples and NBIA families would be better served by placing the samples at the Oregon Health & Science University, home of Dr. Susan Hayflick's lab.

The boards also cited duplication of effort for supporting the

(see *BioBank* on pg. 9)

NBIA research making strides but future funding at a crossroads

NBIA researchers are making strides with mouse models to study brain iron, identification of possible new NBIA genes and studies that could lead to treatments, participants were told at the Fifth International NBIA Disorders Association Family Conference in Indianapolis.



Winifred Williams from St. Louis, Missouri discussing research with Dr. Susan Hayflick from OHSU.

But scientists also said they are deeply worried about supporting their work in the months and years ahead.

"We are facing a critical shortage of funds for our research," said Dr. Susan Hayflick, NBIA's top researcher for the past 17 years. "There are scientists all over the country who are facing closure of their laboratory."

Genetic counselor Allison Gregory who works with Hayflick at the Oregon Health & Science University in Portland said that in the past two years funding "has gotten really tight" from the National Institutes of Health, which supports most of the research by Hayflick and other scientists who study NBIA and other diseases. Established labs like theirs are fearful of running out of money and have been turned down for continued NIH support, Gregory said.

"We want to keep working on this disorder," Gregory said, but without support from other sources, including families, researchers are in danger of closing down projects.

At stake is some strong progress, Hayflick said, "but we have a really long way to go."

Right now, 379 families are in the OHSU NBIA registry, which gives researchers a treasure trove of information about the disorder, including blood and tissue samples to study.

Two genes have been discovered, but scientists have yet to learn why those genes produce iron accumulation in the brain, Hayflick said.

She believes there is enough DNA from samples in the registry to find additional genes. "Many families in this room have disease not caused by either gene. We hope to have promising results this summer," Hayflick said.

She wants the registry to expand and also places high priority on drug development and testing for NBIA patients. More scientists need to be working on the disease, she said.

Dr. Paul Kotzbauer, an assistant professor in the Department of Neurology at Washington University School of Medicine in St. Louis, suggested that working with researchers who study Alzheimer's and Parkinson's disease could be useful, given what he is learning from his mouse models.

Kotzbauer said the models could produce a better understanding of NBIA impairment and guide the development of new treatments. A challenge, however, is that many human neurodegenerative diseases are not easily reproduced in mice. A mouse model he has generated for *PLA2G6* mutations, for example, shows signs of neurodegeneration but does not show iron accumulation. A mouse model for *PANK2* mutations also does not show iron accumulation.



Dr. Paul Kotzbauer from Washington University discusses his research work with mouse models.

For that reason, studies are underway now to combine mutations of the two NBIA genes within the same mouse to get an improved mouse model with which to work.

Dr. Eden Haverfield, assistant director of the University of Chicago Genetic Services Laboratory in Chicago, discussed a study being done there to identify additional mutations in *PANK2* and *PLA2G6* genes not detected by full gene sequencing. These are called intragenic deletions and duplications.

Researchers want to know how common these deletions and

(see research on pg. 7)

Research

(continued from page 6)

duplications are in NBIA disorders and whether they contribute to the disease, Haverfield said. The scientists analyzed 72 patients for *PANK2* deletions or duplications and 12 were positive, while for *PLA2G6*, two of 22 were positive, indicating that intragenic deletions and duplications do contribute to the molecular basis of NBIA.



*Jessica Coates from Birmingham, Alabama
with Dr. Eden Haverfield from the University of Chicago.*

Dr. Ody Sibon, a professor from University Medical Center of Groningen, The Netherlands, is studying the fruit fly to better understand the mechanisms behind certain movement impairments faced by NBIA individuals. Ultimately she wants to know: What



*Dr. Natale Scalise from Rossano, Italy
(President of AISNAF - sister organization in Italy)
and Dr. Ody Sibon from The Netherlands
enjoy the family picnic held on Saturday at the conference.*

therapies might help people with NBIA?

She is testing a compound on the fruit flies to see what impact they have on movement and other disease symptoms, as well as life span.

Finally, Dr. Amande Pauls, a movement disorders specialist at University Hospital of Cologne, Germany, gave preliminary results of a study on deep brain stimulation being led by Professor Lars Timmermann, who could not be present.

The study involved 24 NBIA patients in 15 countries who had DBS, a process in which electrodes are attached to the brain and regulated by a stimulator implanted in the chest to reduce painful involuntary movements also referred to as dystonia. About half showed improvement, Pauls said. The more dystonia the patient had, the greater the improvement, which made a significant difference in the patient's and the family's quality of life, she said.



Dr. Penny Hogarth from OHSU (left) and Dr. Amande Pauls from Germany at the family conference picnic.

In some cases, the painful movements, or dystonia, returned after several months, Pauls said.

Questions remain about when to have DBS done, and the group would like to do a different kind of study, one that starts at the beginning with 20 to 30 patients and follows them forward, rather than looking back on their experience with DBS, Pauls said. If anyone is interested in pursuing DBS, Pauls ask that they contact Timmerman's lab at Movement Disorders and Deep Brain Stimulation, Department of Neurology, University Hospital of Cologne, Kerpener Straße 62, 50924 Cologne, Germany or amande.pauls@uk-koeln.de.

Clinical trial underway on chelation therapy for NBIA



Nancy Sweeters, pediatric nurse practitioner at Children's Hospital and Research Center in Oakland, Calif.

Chelation therapy, which uses chemicals to sweep metals from the body, has attracted the attention of researchers who want to know whether it can safely and effectively remove iron from the brains of NBIA-affected individuals.

Deferiprone is the first iron chelation drug able to cross the blood-brain barrier, and while it is not licensed in the United States, it is approved in 48 countries, including all members of the European Union, and it can be studied

by U.S. researchers, said Nancy Sweeters, a speaker who appeared on a panel at the recent NBIA Disorders Association conference.

Sweeters is a pediatric nurse practitioner at Children's Hospital and Research Center in Oakland, Ca., with Dr. Elliott Vichinsky, who is planning to launch an open phase one study on deferiprone to include NBIA patients.

Chelation therapy is controversial. A study on deferiprone in Canada was halted several years ago because of safety concerns, although some disputed the findings. The drug does have side effects Sweeters said, "It can lower your blood count significantly and you are at great risk for infection," she said. "We don't take it lightly."

The drug also has been used on patients with blood disorders and heart problems. Some studies say the drug has shown promise and have urged further study.

Sweeters said one reason Vichinsky wanted to pursue a trial is because of the positive results in other small studies, including one involving a 60-year-old woman with NBIA who improved on deferiprone. "Based on these data, we went forth, very cautiously trying the deferiprone," Sweeters said. "And it was because we were very comfortable with the drug that we went forward with this."

Veronica Bonfiglio said her NBIA-affected son, Brent, 16, received approval to try the drug as a "compassionate use" in November 2007. Vichinsky oversaw the chelation treatment.

"For us the experience has been incredible," Bonfiglio said. "The small changes have been big changes in our lives."

In February, Brent started to take a few steps on his own, she said. He also is able to sit on his own, something he could not do before. Bonfiglio said Brent has shown improvements in speech and

handwriting as well.

"I don't know how long this is going to last," Bonfiglio said. "Sometimes I feel like Cinderella waiting for midnight."



Gaetano, Brent, Julian and Veronica Bonfiglio visit the Indianapolis Motor Speedway while in town for the family conference.

Dr. Penny Hogarth, a neurologist and associate professor at the Oregon Health & Science University, told parents to "proceed with caution (and) proceed thoughtfully."

"You can't make a direct translation from one disease to another," she said. No one knows how this drug might affect patients with NBIA and whether some might be harmed or even die from it, she said.

Also, in the ataxia trial, of the 13 patients who were studied, four became very ill and one was paralyzed with Guillain-Barré syndrome, Hogarth said. "We all need to be careful how we interpret the results," she said.

At the same time, Hogarth praised the way Vichinsky and Sweeters are doing the deferiprone study.

"The way they are going about it is exactly the way it needs to be done: thoughtfully, carefully, dealing with the bloody FDA, the way the FDA likes to be treated, and very carefully assessing the outcomes," Hogarth said.

She added that she was not trying to dissuade patients from enrolling but encouraged them to consider all of the information. In the case of NBIA, "we don't know...if iron is causing the problem," Hogarth said.

Sweeters said the study is "not for everyone."

"This is just a piece of the puzzle," Sweeters said. "We are trying not to promise anything of a miracle. We are trying to say that we are willing to help you with this step because of our experience with deferiprone."

(see chelation on pg. 9)

Chelation

(continued from page 8)

As we went to print we learned that the FDA has agreed to a 6 month Phase I, double-blind, placebo controlled study of deferiprone in approximately 20-30 NBIA patients. Two-thirds will get deferiprone and one-third will get a placebo. Dosage will be 20-30 mg per kg daily.

Is gene therapy a possibility for NBIA?



Dr. Ajay Perumbeti from Cincinnati Children's Hospital Medical Center discussed gene therapy on Sunday morning at the family conference.

While gene therapy is experimental today, it might one day be used to treat NBIA and a host of other diseases, said Dr. Ajay Perumbeti, a fourth year fellow in hematology/oncology at Cincinnati Children's Hospital Medical Center.

Gene therapy involves treating disease by introducing a new gene into the body or replacing a mutated gene with a healthy one, said Perumbeti, who spoke during the Fifth International NBIA Disorders Association Family Conference.

Researchers have been studying gene therapy for several decades, but it still is considered experimental. Although there was evidence for great potential for clinical application of gene therapy in the 1980s, the results of clinical trials were initially disappointing, Perumbeti said, because we did not know how to optimally design and deliver effective genes that would not be silenced. There has been much progress in gene engineering and delivery since then.

These days, researchers are still finding it easier to add a gene than replace one, but safety remains an issue. "You can potentially cause cancer by inserting a gene," Perumbeti said, but researchers are currently developing and testing ways to reduce this risk.

The Food and Drug Administration has not approved the sale of any gene therapy products, and progress has been slower than anticipated since the first clinical trial in 1990. A major setback occurred in 1999 when 18-year-old Jesse Gelsinger died four days after being injected with a genetically altered virus designed to treat his liver disorder, secondary to an immune response.

Another blow came in 2003, when FDA halted all trials that use retroviral vectors for inserting genes into bone marrow stem cells. That occurred because a second child treated in a French gene therapy trial for severe combined immune deficiency (SCID), a lethal immune system disease, had developed leukemia.

Although this was evidence that safety concerns were more than hypothetical, it has also become clear that genetic therapy has now cured many patients with SCID. The latest report on long term results of gene therapy for ADA SCID, a type of SCID, showed resounding success and was published in the New England Journal of Medicine in January 2009.

There are encouraging signs for neurological diseases, too. Gene therapy research with Parkinson's and Alzheimer's diseases is progressing, including ongoing clinical trials. Delivering antibodies to the brains of Alzheimer's patients showed signs of disrupting plaque, Perumbeti said.

An issue is how to deliver genes, particularly ones that need to cross to the blood-brain barrier. One possible method is to direct injection into the brain. Others are developing gene carriers that will penetrate the blood brain barrier.

With the discovery of two NBIA genes, there is every reason to think gene therapy could be a promising strategy for NBIA, he said. The big advantage that NBIA has is that there has been some success in characterizing the major genes that cause the diseases and developing models to test treatments by the Hayflick Laboratory and others.

"Gene therapy holds much promise for amelioration, correction and cure for diseases, including NBIA," Perumbeti said. "But first we must find candidate genes and test them in good disease models."

BioBank

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move.

BioBank costs in 2004 were expected to be \$5,000 to \$10,000 a year and were supported with a \$50,000 grant the board received specifically for this purpose. In 2007 the BioBank moved to another facility and charges began to rise. Costs had reached about \$15,500 per year for the storage of 107 samples from NBIA families.

Because so many of the families also had samples at the OHSU registry, there was an overlap. Both banks made samples available to other NBIA researchers, and because that will continue, research will not be affected by the change.

Hayflick has pledged that in the event her lab should close, any NBIA DNA stored in her lab would be maintained at OHSU and would remain available to other NBIA researchers.

Brain tissue donations using the facilities and expertise at the National Institutes of Health will continue to be a priority for the organization. Matthew Hodgson, who directed the association's BioBank program, has agreed to continue as a volunteer to help coordinate this vital need for research.

A Special Thanks to the Fifth International NBIA Disorders Association Family Conference Sponsors

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Ability Magazine ran a full page ad for NBIA Disorders Association in their April 2009 issue in exchange for being named our media sponsor for the family conference. We also ran a full page ad for them and their non-profit organization, Ability Awareness, in our conference program, distributed their magazine at the conference, acknowledged them at our Web site and in this newsletter.

Family Conference Scholarship Contributors

NBIA Disorders Association would like to thank the following individuals, for it is through their generosity that we were able to offer partial scholarships for families and individuals to attend who otherwise would have been unable to participate in the Fifth International NBIA Disorders Association Family Conference.

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Diane Murdock from Indianapolis, Indiana and Vicky Stapleton from Cincinnati, Ohio accepted appreciation certificates at the family conference for their sponsorship and volunteer support.

F amily Conference P ictures



Participants at the Fifth International NBIA Disorders Association Family Conference in Indianapolis, Indiana May 29-31.



Drew Karakourtis from Austin, Texas is ready for rain.



Cameron Meade from Dimondale, Michigan meets a pet therapy dog in the care room at the conference.



Brent Bonfiglio, Macey Murdock and Victoria Saraceno get wrapped in toilet paper by their teammates in a picnic game.

NBIA association hires first paid staff member

By Patricia Wood



Luann Rein, who lives in San Diego, Calif. is the organization's first Executive Director.

After 13 years, the NBIA Disorders Association had hired its first paid staff member, Executive Director Luann Rein.

Rein, who lives in San Diego, home base of the organization, will take over many of the day-to-day operations. While this is a part-time position, we hope to someday make it full-time.

This is an important milestone for our previously all-volunteer organization. It is both a positive and necessary step that the board believes will move our organization forward.

Rein has a rich background of volunteer non-profit experience. While volunteering for the Make-a-Wish Foundation of San Diego, she helped raise donation levels from \$6,000 annually in 1992 to \$268,000 a year in 2002 by training a speakers bureau to give talks in the business community about designating donations to Make-a-Wish.

We plan to tap her fundraising expertise to assist you and others who care about NBIA to do fundraising in your communities. She also will be involved in helping us to expand our online fundraising opportunities through sites such as Facebook and Firstgiving.

Rein's other tasks include helping to produce the newsletter three times a year and assisting in revamping our Web site, as well as outreach to businesses for support.

Rein has a bachelor's degree in psychology from the University of California, San Diego. She owned a gourmet pastry shop and was the lead pastry chef for 22 years, has taught improvisational theater to schoolchildren and was the general manager of a multi-media art studio.

You can reach her at Irein@NBIAdisorders.org.

Mother of three with NBIA says she's 'too blessed to be stressed'

By Cheryl Lamos

I recently opened one of those e-mails that goes around that had a picture of rolling green hills and the caption, "I am too blessed to be stressed." Ever since it has been on my mind.



Cheryl Lamos and her daughter, Barbara Belcher, from Rensselaer, New York, at the family conference in May.

I was blessed with four beautiful children who grew into wonderful adults. They would make any mother proud. That's not to say I haven't been stressed. Since 1995 I have struggled with the heartbreaking diagnosis of NBIA in three of my four children. And my son Ben, the one who doesn't have

NBIA, had a seizure disorder between the ages of 8 and 18. Fortunately, Ben, now 24, has been seizure-free for six years.

There has been tragedy, too. My son Bruce, at the age of 26, developed deep vein thrombosis in his leg after breaking his arm. A piece of the blood clot broke off and traveled to his lungs, killing him. I like to say he moved to heaven before the rest of us because he always liked being first at everything.

My daughters, Becky, 32, and Barbara, 29, love to laugh and have been wonderful daughters.

As a single parent and working full-time as a nurse, I recently allowed myself to become too wrapped up in worrying about work. I was extremely stressed and also isolated myself from friends and family. I was depressed about how unfair life can be, feeling sorry for myself and couldn't sleep. I think you get the picture.

I ended up in the ER one evening with a panic attack. My doctor advised me to rest, and I took a month off work. I learned some valuable lessons and finally worked through the guilt I carried about

*(see **blessed** on pg. 13)*

Blessed

(continued from page 12)

passing on a defective gene to three of my beautiful children.

I decided I needed a less stressful job so I could be home more with Barbara at night. No more working from noon to 10 p.m. four days a week. I transferred to a new floor at the hospital and now work 10 a.m. to 6 p.m., five days a week.

I reset my priorities and realized it was my choice to be miserable or happy. Do I want to feel sorry for myself or realize all of my blessings and enjoy my life? I worked through my stress and now am much happier than I have ever been. I try to enjoy each day.

Although Becky has been in a nursing home for the last year, she still has a wonderful smile, especially when Barb and I play her favorite Beatles songs and sing along. Yes, Bruce did leave us sooner than I liked but I believe in God and heaven. I know I will see him again. Yes, Barbara can be annoying at times but she wakes up every day with a smile on her face. Yes, I miss Ben who is on the other side of the world teaching English in Japan, but I am so proud of him. Since his brother passed on, he values his life and college education much more.

What I am trying to say is that I am truly very blessed and maybe I had to go through some dark valleys before I realized it. I learned that my friends and family can't help me or support me if I don't let them know what I am going through. At age 54, I am still learning and I don't want to stop.



*Cheryl Lamos with her four children in 1981.
Standing front row: Barbara, Bruce and Becky.
Ben is in his mother's arms.*

I almost didn't go to the NBIA conference, but I am so glad I did. I am glad for the support we can be to each other, and while it is hard opening yourself up to strangers, it is so worth it. We need to know we are not alone in this struggle and to remember to take care of ourselves.

We have been blessed with beautiful, special children. Let's not be stressed, but blessed.

**For more information on this family's story, go to Family Stories on the Web site under the Family Support link. They were featured in April 2004.*



*Aubrey D. Anielle L. Ockard
November 7, 1997 - March 20, 2009*

Hello NBIA family,

With a heavy heart I notify you that we lost our daughter, Aubrey, to septic pneumonia and respiratory failure on March 20. Aubrey was a special soul lent to us by God for a short time, but she left lasting impressions on everyone she touched in her 11 years.

Aubrey was a courageous child who got right back up whenever she fell and kept going full steam. From the moment she was born we knew she was touched by God. She was our angel on earth.

Although Aubrey was taught at home, she was adopted by her fourth-grade peers and counted many friends among them. They visited with her weekly for lunch, reading to her and entertaining her. Her teachers and peers went out of their way to keep Aubrey connected and brought tremendous joy to her life.

Aubrey enjoyed watching Blues Clues, spending time with her "Pa" and lying on the floor with her nurse, Susan, and her dog, Dexter. Aubrey spent her 9th birthday at Disneyland through Make-A-Wish and loved the princesses and the Dumbo ride.

Aubrey started out as a normal child but quickly developed difficulties with her milestones and received many different diagnoses before being confirmed with PKAN two years ago. She quickly lost all of her independence, but she took it all in stride. She was very brave.

Since Aubrey's passing there have been many trees planted in her honor in addition to a new award named after Aubrey at her school. It will be given each year to a fourth grader showing caring and courage.

(see Aubrey on pg. 14)

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

Barbara & Becky Belcher
Cheryl Lamos
Doug & Joy Lamos

April & Tracie Flinn
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Ronald & Anne Walker
Thomas & Mary Colleen Wood
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Ernestine & James Younger

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Alyssa & Sabrina Barbiero & Family
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Bruce Belcher
Cheryl Lamos

Madison Frederick
Randolf & Doris Frederick

Joyce Knott
Bernie & Judy Roberts

Aubrey Lockard
Shelley Barrett
Shirley Cholh
Dennis & Bridget Perkis

Wade Patton
Paul & Janet Buhay

Richie Roberts
Bernie & Judy Roberts

Ken Stromsta
Linda & Rick Reavis
Charles & Shelley Sprackling
Al & Pam Stromsta

Aubrey

(continued from page 13)

Aubrey is dearly loved and missed by her parents, grandpa, Susan, her sister and two brothers, in addition to her extended family of peers, teachers, therapist and all of the loving and giving people who came into our lives because of Aubrey.



Devon Fletcher, mother to Aubrey Lockard and Pam & Al Stromsta, parents of Kenny, were on hand for the memorial tribute at the family conference picnic.

MESSAGE FROM THE PRESIDENT



Patty Wood

When Dr. Susan Hayflick told me that her all-important federal grant was denied and her lab at the Oregon Health & Science University would probably have to close by the end of the year, I still could not believe it. I had known this was possible, but reading the message in her e-mail shook me to the core.

This could not be happening to the person who has been my friend and my hope all of these years.

My path has been tied to Dr. Hayflick's since I met her in 1996. I flew from San Diego to Oregon immediately after learning a researcher was interested in the disease that was robbing my daughter of her abilities.

We decided to work together that day to create an organization that provided more information than the small paragraph I found in the Mayo Clinic medical library hours after Kimbi was diagnosed in 1989. That paragraph has proven itself to be full of inaccuracies.

Dr. Hayflick and I wanted to unite NBIA families and promote research. Thirteen years later, we can say that much progress has been made, and newly diagnosed families have so many more options than my family had in 1989. But there is still much to be done.

Dr. Hayflick is the major reason I have hope that a treatment will be found to help many of those affected by NBIA. She is the reason I believe that a cure is possible.

A few weeks ago, I started thinking about ways to raise \$250,000 quickly to keep her lab open for the next two years and the lottery came to mind. I've seldom played but off I went to the 7-11 convenience store to buy my chance for a miracle. I asked the clerk many questions — how many numbers do I pick, how much is the jackpot, how soon is the drawing? I could see by the look in his eyes he was contemplating what crime I had committed and why I was so desperate to win.

I thought surely God would see this was a good cause and intervene so that my numbers would come up. It didn't happen, but I am still buying tickets — just in case.

Now, I am reconciled to thinking that it seems we are going to have to make this miracle happen the old-fashioned way — by working hard and hoping that somehow, some way, everything will work out. I simply can't image another alternative.

So I'm asking everyone who receives this newsletter — or anyone

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.

Angela Bonfiglio Allen	Franco Mau
Gary Belcher	Eldon & Sandra Nelson
Jeffrey & Kimberly Botsford	Neil O'Shea
Cheryl Brooks	Quest Diagnostics
Don Carter	Quintiles, Inc.
Michael Cohn	Jerri Rednour
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Frances Klosterman	Dr. Rayburn & Ardell Skoglund
Gerald Kneisi	Jerry & Victoria Weiner
Robert & Patricia Kromdieck	Leslie & Steven Zent

Missed the conference?

We have a set of 5 DVD's that cover many of the sessions at our Fifth NBIA Disorders Association Family Conference.

Thanks to the generosity of Cincinnati Children's Hospital who loaned us video equipment and the expertise of Ronda Inman who volunteered her time, we can offer these sets to anyone interested for just \$12.

(plus shipping if outside the U.S.)

If interested, please contact Patty Wood at pwood@NBIAdisorders.org or call at (619) 588-2315.

Topics covered: Welcome by Patricia Wood, Keynote Speaker Brian Denger - Parent Advocacy, Updates from Program Directors: Mike Cohn, Sandy Leap, and Megan Thomas, NBIA Families Info Sharing, Fundraising and Federal Support, Assistive Technology, What's New in NBIA Research? Iron Chelation Treatment (Deferiprone)

who is interested in our cause — to come forward and help us make the Hayflick Lab Campaign a success. Please pass this information on to your family, friends and associates and ask them to help us. You can make a difference.

In desperate times, I think it's helpful to recall the words of anthropologist Margaret Mead. She said, "Never doubt that a small group of thoughtful, committed citizens can change the world; indeed, it's the only thing that ever has."



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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

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from discovery to cure

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