

The compound pantethine shows promise as possible future treatment for NBIA

By Dr. Ody Sibon

With the help of a 2008 grant from the NBIA Disorders Association, my research group collaborated with the group of Dr. Susan Hayflick in Oregon to see whether the compound pantethine could be a treatment for PKAN, a form of NBIA.

We were encouraged by what we found in our investigation using fruitflies, although more research is needed before we could test this potential in humans.

We previously demonstrated that the fruitfly (*Drosophila*) model for PKAN could be used to better understand PKAN at the cellular level.

As many of you know, the use of fruitflies to understand diseases is not unique for PKAN and they are currently being used for other neurodegenerative diseases such as Parkinson's and Huntington's.

The PKAN fruitflies carry a mutation in the *Drosophila* pantothenate kinase gene, and like in humans, that gene encodes for an enzyme that is required for the synthesis of Coenzyme A.

(see *pantethine* on pg. 3)



Dr. Ody Sibon from the University Medical Center Groningen in The Netherlands works with fruitflies in her research lab.

Clinical trial on deferiprone seeking patients; hoping for April 2011 launch

By Veronica Bonfiglio

A long-awaited clinical trial to test deferiprone as a safe treatment for NBIA is seeking patients interested in participating at Children's Hospital and Research Center in Oakland, Calif.

Dr. Elliott Vichinsky, director of the hospital's hematology/oncology department, and his nurse associate, Nancy Sweeters, hope to enroll 20 to 30 patients in the trial. Those patients would need to make four visits to Oakland. The visits will be at study start, 3 months, 6 months and 1 year.



Dr. Vichinsky and his team will be submitting the grant to the National Institute of Neurological Disorders and Stroke (NINDS) which is a branch of the National Institute of Health (NIH) for a July 14 deadline. In this grant they

Dr. Elliott Vichinsky, from Children's Hospital and Research Center in Oakland, Calif. hopes to launch clinical trial on deferiprone.

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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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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.

Two-day symposium in DC area this October chance for NA and NBIA scientists to gather and collaborate

Scientists from around the world will gather in the Washington D.C., area this October to discuss research priorities and ways they can collaborate on NBIA and another rare neurological disorder, neuroacanthocytosis, also known as NA, which also is characterized by movement disorders and seizures.

At the two-day meeting, invited researchers will discuss what is currently known about NA and NBIA at the cell level. The Brain, Blood and Iron: Joint International Symposium on Neuroacanthocytosis and Neurodegeneration with Brain Iron Accumulation has been set for Oct. 1 and 2 at the Doubletree Hotel in Bethesda, Md.

Scientists hope to determine research priorities for both disorders, decide what resources are needed to further that work and discuss ways to generate collaborations between those working in related disciplines.

While the talks will be highly technical, NBIA families are invited to attend the symposium and will only be asked to pay a fee to cover their food costs. The Saturday afternoon session discussing where NBIA research is headed may be of particular interest to families. Any individuals who attend are invited to stay for Saturday night dinner and to meet the researchers. A \$50 fee covers dinner costs.

For a full description of the symposium program, see the home page of the website for the Advocacy for Neuroacanthocytosis Patients at www.naadvocacy.org. Also, contact Patty Wood at pwood@NBIAdisorders if you are interested in attending any part of the symposium.

Pantethine

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The enzyme is critical to metabolism and is required to convert vitamin B₅ into Coenzyme A in humans and in fruitflies.

The mutant flies show neurodegeneration, have impaired locomotor function and have a short life span. PKAN fruitflies also show abnormalities in specific cellular structures called "mitochondria."

Fruitfly models are also useful to test whether compounds have some rescuing potential for specific diseases. My group had preliminary data indicating that the pantethine compound had a protective effect on the PKAN fruitflies.

So, with the NBIA grant, we looked further at the rescuing potential of pantethine. First we showed that Coenzyme A levels in PKAN fruitflies were extremely low. Next we showed that levels of Coenzyme A increased when pantethine was added to the food of the PKAN fruitflies. Restoration of Coenzyme A coincided with rescue of the mitochondria, slowed neurodegeneration and increased locomotor function. It also extended the life span of the fruitflies. These results show that in fruitflies pantethine can serve as an alternative source to generate Coenzyme A when the pantothenate kinase enzyme is defective.

Our research also showed that impaired function of pantothenate kinase 2 in specific human cultured cells also resulted in abnormalities of the mitochondria. These abnormalities were rescued when pantethine was added to the medium.

With these promising results, there is a suggestion that pantethine may serve as a basis to develop a therapy for PKAN. Although a limited amount of clinical studies in which pantethine was used to treat patients with hyperlipoproteinemia have been done by other groups, further research is required because pantethine is not a Food and Drug Administration-approved drug and its effects in humans are not yet clear.

Currently, more studies are being performed in various human cell lines and in mice on pantethine's potential for treating PKAN, and our group's work in this area will continue.

This study was recently published in the April issue of the scientific journal Proceedings of the National Academy of Sciences of the United States of America (PNAS).

Deferiprone

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require that the group shows there is interest in the patient population to go forward with the study. There is no personal information that is sent into NINDS — just age, gender and state of residence.

If the grant is accepted, the trial would launch in April 2011.

ApoPharma, the maker of deferiprone, which is an iron chelator, will provide the medication and placebo for the study. However, ApoPharma is not funding the study. Vichinsky and his team originally thought that might happen, but the company is looking at many other studies involving diseases with brain iron.

Consequently, Vichinsky is seeking a \$1 million grant from the National Institutes of Health. Although the NIH did not fund his application last year, Vichinsky believes his chances this year are improved. He received Investigational New Drug (IND) approval from the Food and Drug Administration in December 2009, a requirement before a study can begin. This approval was based on a placebo-controlled study being done, a change from the initial application. "Having the IND should put us ahead of the pack," Sweeters said.

Anyone who is interested in taking part in the study should contact Sweeters, and those who want to show support for the grant through NIH's National Institute of Neurological Disorders and Stroke are asked to call or e-mail the agency (See accompanying box on pg. 6 for information on how to apply for the study and/or support the grant.) As a patient advocacy group, NBIA Disorders Association is supporting the application and preparing a letter families can sign, especially those interested in taking part in this study.

Vichinsky said grant applications will be peer-reviewed this fall and he hopes to hear by November if the work will be funded beginning next April.

Deferiprone is an iron-removing drug able to cross the brain blood barrier. In the study participants would be taking deferiprone orally.

Deferiprone was approved in 1999 by the European drug regulatory authority and can be freely used in 48 countries. But it is still considered experimental in the U.S. and is tightly regulated, although it has been used by Vichinsky and others to treat complications of the blood disease thalassemia since 2006.

In the past, other iron chelators had been tried with NBIA patients but those attempts failed because individuals would become anemic and treatment had to be stopped. A 2007 French study on Friedreich's Ataxia (FA), another disorder with excess iron in the brain, found that deferiprone was effective in sweeping out the iron

and improving symptoms. That caused some NBIA families to wonder whether the drug could also help their loved ones.

Vichinsky has been using the drug for thalassemia patients since 2006, and in November 2007, my son Brent Bonfiglio of Fremont, Calif., became the first U.S. child to receive deferiprone for NBIA in an FDA-approved compassionate-use clinical trial under the direction of Vichinsky. That protocol allows an experimental drug to be used on a case-by-case basis on very sick individuals who have no other options. He now cares for two other NBIA children, besides Brent, with deferiprone.

Brent currently receives 35mg/kg/day. Vichinsky said he would start doses in the clinical trial at 20 mg/kg and go no higher than 30 mg/kg a day.

I saw Brent, now 17, gradually improve from the drug. Sometimes I would notice something very clearly, like Brent being able to sit without any back support; other times it was more subtle. Regardless, I believe deferiprone put the brakes on NBIA and that my son is better today than he was three years ago.



Brent Bonfiglio, 17, from Fremont, Calif., shows his ability to sit without back support, a skill he has regained since taking deferiprone.

Adam Tifone, 19, of Gibsonia, Penn., has been on deferiprone for two years. His dosage started at slightly less than 20 mg/kg/day for the first eight months, according to his father, Rick Tifone. He was stable during that period, but his family saw a noticeable

(see *deferiprone* on pg. 5)

Deferiprone

(continued from pg. 4)

improvement in his balance when the dosage was boosted to 26 mg/kg/day, Rick Tifone said.

"The balance improvement lasted until early 2010," Rick Tifone said in late May. "Three weeks ago his dosage was increased to 35/mg/day. We haven't yet seen any noticeable improvement at this higher dosage. He has not had any side effects from the drug in the two years he has been on it. Overall, I would say his condition is about the same as it was two years ago, which given the degenerative nature of this disease, the deferiprone treatment has been a success."



Adam Tifone, 19, of Gibsonia, Penn., has been taking deferiprone for two years without any side effects.

I know of about 10 other NBIA patients undergoing treatment in the U.S., Cuba, the UK and Italy where Dr. Gian Luca Forni of Centro della Microcitemia, Ospedale Galliera, Genoa was the first to treat NBIA patients with deferiprone.

Results to date have been encouraging.

Last year, nine PKAN patients in Italy completed a six-month trial with Drs. Bertini and Nardocci using a dosage of 25 mg/kg/day and

(see deferiprone on pg. 6)

Enrollment Criteria for Deferiprone Trial

Based on preliminary enrollment criteria, deferiprone study participants must:

- * Have a confirmed diagnosis of NBIA.
- * Have favorable results from the initial screening that includes comprehensive blood work and a medical evaluation.
- * Be at least 8 years old.
- * Have the ability to travel to Oakland, Calif., four times during the study. Researchers hope to have money to reimburse for travel expenses.
- * Agree to a weekly blood draw for safety.
- * Have had a stable setting for at least two months before the study begins, if the person has a deep brain stimulator.
- * Have had a baclofen pump for at least two months before the start of the study. No pump is fine.
- * Have video recordings, along with physical therapy and occupational therapy evaluations at every visit to Oakland.
- * Have brain MRIs.
- * See a local neurologist every other month.

To find out more about applying to take part in the study

Contact Nancy Sweeters, associate nurse to Dr. Elliott Vichinsky, at nsweeters@mail.cho.org.

Also notify NBIA Board Member Veronica Bonfiglio at veronica_bonfiglio@sbcglobal.net.

To support the grant application

Contact the NIH's National Institute of Neurological Disorders and Stroke to express the urgency for this clinical trial by calling, writing or e-mailing to:

Dr. Katrina Gwinn
 Program Director, Extramural Research Program
 NIH/NINDS
 Neuroscience Center, Room 2143
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Deferiprone

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tolerated deferiprone well. Publication of those results are still pending.

It's possible that some patients may not benefit, and deferiprone has well-known side effects. Safety data has been collected from about 1,000 patients, including 500 who have been on the drug for at least two years.

The most common side effects are reddish discoloration of the urine, nausea, vomiting, joint pain, abdominal pain and an increase in ALT, an enzyme that measures liver function. In general, those reactions were mild to moderate and generally go away without stopping deferiprone.

A major side effect is lowered white blood cell count, known as "neutropenia" or "agranulocytosis," depending on the severity. That side effect was seen in about 1 percent of patients and went away when deferiprone was stopped. A low white cell count puts you at risk for serious infection and must be promptly treated.

"This is hope for a tragic, hopeless disease," Vichinsky said, "If deferiprone can get enough iron out of the brain, we believe it can reverse a significant part of the disease."

At the 2009 NBIA 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" if they choose deferiprone. 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. "Deferiprone may treat it, but it doesn't cure it."

She agreed, however, that more research is needed for what some consider a potentially promising treatment for NBIA. And if patients are going to try it, they might want to consider doing it under the care of Vichinsky, she said. "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.

Dr. Susan Hayflick, NBIA's premier researcher in Oregon is involved with the trial and has a close partnership with Vichinsky.

"This is hope for a tragic, hopeless disease," Vichinsky said, "If deferiprone can get enough iron out of the brain, we believe it can reverse a significant part of the disease."

If positive results from the study are found, then all patients participating in the clinical trial will be eligible to receive deferiprone from ApoPharma afterward. Even if the trial is successful, it still would take several years for the drug to go through the FDA's approval process and become widely available to those who do not participate in the clinical trial.

Spanish-language listserv now up and ready to use

By Veronica Bonfiglio



Veronica Bonfiglio from Fremont, Calif. is a member of the association's board of trustees and moderator for our new Spanish listserv.

Hola! NBIA Disorders Association's listserv is now bilingual.

As a native Spanish-speaker, I have been serving as the association's unofficial ambassador for Spanish-speaking families who contact our organization desperately seeking information. I have been corresponding with families from Spain, Mexico, Cuba, Dominican Republic, Chile and Argentina.

Because so many of them speak no English, they miss out on the wonderful info exchange that is possible through the listserv. I wondered, why not start a listserv group in Spanish then? President Patty Wood agreed. And now, here we are.

As the moderator for this new group I make sure that any important, new issues are translated into Spanish. I also see to it that any significant issues that arise from the Spanish-speaking families are communicated to the English listserv. The Spanish listserv is proving to be a wonderful new outreach program for us.

If you speak Spanish or would like to refresh your Spanish language skills, we'd love to have you join this new listserv group. If interested, please e-mail me at veronica_bonfiglio@sbcglobal.net.

Mother who lost one son to NBIA has hope for the second son

By Joan Kissling

I knew when Dr. William Yang was calling in June 2006 it was going to be bad news. As he told me the results of the most recent MRI, about finding iron build up in my child, I started sobbing.

As I was fighting for breath and trying to thank him for calling, I was ready to hang up. He pleaded with me not to. He had found a medication that might help. I listened to what he believed was my 11-year-old son's only chance.

We had already lost our first son to NBIA in 1999 at the age of 19; we could not bear to lose another.

I listened to what the doctor said that day as he described the drug deferiprone. We decided we were not willing to wait years for the Food and Drug Administration to approve its use, so we went out of the country to get the medication. We found a doctor brave enough to give us a prescription and the doctors here were willing to monitor my son's blood work to make sure we did him no harm.

Deferiprone's side effects were known and the doctors knew what to watch for. My son, Angel-Andres, has been on it ever since.

Every child is different but I cannot help but to compare my two sons. At age 16, my first son was wheelchair-bound and was extremely hard to understand. My second son is still walking, talking and, at first glance, appears to be a normal healthy teenage boy.

I am convinced that this medication has slowed down the progression of the disease and actually improved his speech and balance. He was starting to stutter for a while but that has gone away.

We parents are told by doctors that there is no treatment, that we need to wait for more research. There are no other options. I will always be grateful to Dr. Yang, now deceased, who was willing to take an educated risk that most doctors would not do.

Angel-Andres is now 16 and rarely gets sick. If he does get a cold or cough, I take him off the medication until he is well again. This medication can have a rare but serious side effect of lowering the white blood cell count. White blood cells are essential in fighting off disease. The last thing any parent wants is for their child to die of a common cold. Angel-Andres has had no noticeable side effects. His white blood cell count has been excellent.



Angel-Andres Diaz, 16, from Las Vegas, Nevada has NBIA and has been on deferiprone since 2006.

Every child is different but I cannot help but to compare my two sons. At age 16, my first son was wheelchair-bound and was extremely hard to understand. My second son is still walking, talking and, at first glance, appears to be a normal healthy teenage boy.

He gets blood work done regularly and it is monitored by a neurologist and a pediatrician. On one occasion his liver enzymes spiked but went back to normal within one week. The doctors do not think it was the medication.

At this point, we realize our story is just a testimonial, although it is very interesting to other desperate parents and to the doctors. We need valid clinical trials to determine if this medication truly helps. And we have to find funding for Dr. Elliott Vichinsky's clinical trial in California.

Congressman asks for federal funding of \$2.5 million for NBIA research

By Dr. Mark Karakourtis

March was an exciting time for our family. I went along with our son Drew's class in Austin, Texas to Washington, D.C., and Drew and I were hoping to meet with our congressmen, Republican Lamar Smith and Democrat Lloyd Doggett while there. We got two meetings — and more than we dreamed possible.

It didn't start out all that promising, however.

On our first full day in D.C., Drew's class was scheduled to meet with Doggett, who serves on the House Budget and Ways and Means Committees. Unfortunately, Drew woke up vomiting and with a fever. He was confined to the hotel for most of the day.



Dr. Mark Karakourtis and his son Drew from Austin, Texas, meet with Congressman Lloyd Doggett. on a visit to Washington D.C. in March.

And unfortunately for his class, the congressman had to cancel because of meetings. However, Doggett did send his senior legislative assistant, Ezra Levin, to meet the class and I contacted Levin later that day to set up a meeting. He was not optimistic that we would get to meet with his boss at any time during our stay because of scheduled Appropriations Committee meetings.

But persistence paid off.

After several e-mails and phone calls Doggett graciously agreed to meet with us during a break the next day. We first met with Levin then with Doggett for about 20 minutes on Independence Avenue right across from the Capitol. We were able to tell Drew's story and talk about his issues with NBIA. We also spoke of the need for funding and research.

The congressman authored a letter which was sent to the House Appropriations Committee on May 11, requesting \$2.5 million in the upcoming fiscal year for NBIA and rare disease

research from the National Institutes of Health. Of course, the request would have to go through the congressional budget process, but it's a start.

On our last full day in D.C. we also were able to meet with Smith, the ranking member of the House Judiciary Committee as well as a member of the Science and Technology and Homeland Security committees.

We were scheduled to only attend a "meet and greet" with other lobbyists and constituents, and I was concerned that it would be difficult to spend any meaningful time with the congressman. We first met with Ashlee Vinyard, his administrative director, and had a productive meeting in which we once again discussed NBIA and the lack of research funding. Drew was a trouper.

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Congressmen

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As we were waiting for the "meet and greet" to start, Smith invited us into his office and we were able to meet one-on-one for about a half hour. We talked about Drew and NBIA but also spent considerable time talking about the lack of research funding. Smith was moved and committed to writing a letter to the NIH requesting funding specifically for the research of NBIA.

On a personal note, the congressman invited us to go with him to the Capitol as he voted on the House floor. Drew answered several of the Congressman's questions as we rode the subway to the Capitol. We even got a private tour of the Capitol and were informed later that Smith wrote the funding request letter and of his intent to circulate it among the entire Texas delegation asking for co-signers.

Smith's office told us the letter "will be sent to NIH and will request a commitment to fund research on NBIA. It discusses the impact of your meeting and the Congressman's understanding of NBIA and neurodegenerative disorders. It also discusses the lack of NIH funding for research for NBIA and the challenges facing the research labs. We will let you know when we send the letter to NIH and all of the co-signers. Thank you very much for the detailed information."

We are encouraged by the response we received from our congressmen and are greatly appreciative to them and their staff for the time they took to meet with us and make us feel welcome.

Now is a good time to do the family's estate planning

*By Megan Thomas**

We are all used to planning every day, whether it's to take care of our children, our families, our homes or issues at work. We make plans so that when we are not at home or at work, someone responsible will carry out our instructions.

The same concept applies to estate planning. What's that? Fundamentally, estate planning is good stewardship of your financial and personal resources so that you have a plan in place to care for your family. It may involve a will or revocable trust, appropriate insurance, planning with retirement assets, or some other kind of financial plan.

When a child with special needs is a possible beneficiary, all adult family members should be involved, including parents, grandparents and older siblings. Their estate plans should supplement but not

interfere with benefits the child may be entitled to receive. Such mistakes can require expensive court proceedings to correct.

Sadly, a majority of Americans die without a valid will. In some respects, we all have a will — your state makes one for you if you die without preparing one. But state laws that govern how your property passes without a will often fail to meet the needs of surviving family members — especially if a member of your family has special considerations. They also fail to honor your charitable intentions — another part of estate planning.

Achieving the goals of research and a cure for NBIA depends on financial support today and in the future. A bequest under your will or from your revocable trust is the simplest way to make a gift of future support to 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."

Please note: The information contained in this article is not intended to be legal or tax planning advice. Please consult your attorney or tax advisor for advice concerning your estate plan.

*Megan Thomas is director of the NBIA Disorders Association's Planned Giving Program and a lawyer.



Megan Thomas and her daughter Sonja Olson from Princeton, New Jersey.

NBIADA's 2009 fiscal year one for the record books

By Patricia Wood

Despite the tough national economy, the NBIA Disorders Association took in a record amount of donations in 2009: \$230,527. That was nearly three times as much as the \$84,000 the organization raised in 2008.

More than half of the donations in 2009, \$127,076, went to the Hayflick Lab Campaign, an emergency effort to keep open the lab of our premier researcher, Dr. Susan Hayflick. Also, it should be noted that 2008 donations were 23 percent below what was raised in 2007.

In 2009, the organization attracted 1,300 donors with the average donor contributing \$180. Half of all donors gave \$50 or more and 44 donors gave \$1,000 or more. Our largest single donation was \$10,000 from Global Knowledge of Cary, N.C., the employer of NBIA parent Mike Fox of Raleigh, N.C.

Our expenses for the year totaled \$243,792. As the pie chart shows, the NBIA Disorders Association spends its money in the following ways:

**NBIA Disorders Association
2009 Expenditures**



- Administration (3%)
- Internet presence (4%)
- Fundraising (5%)
- Scientific Workshop/Family Conference (9%)
- Newsletter (10%)
- Research (70%)

***Administrative expenses.** These include postage, office supplies and other such costs. Other management expenses are for an independent CPA financial review and for state registration fees, which are required expenses when doing fundraising in various states.

***Our Internet presence.** Our website is continuously updated with the latest research information, organization news, and it allows us to accept donations online. This is a very important way for us to educate the public and raise awareness of NBIA. We also have a group and cause page on Facebook and are branching out to include other forms of social media under Luann Rein's direction.

***Fundraising.** Our Hayflick Lab Campaign had some fundraising expenses connected to it and we also put a portion of Luann Rein's part-time salary under this heading. Rein's position began in March 2009 and her title recently changed from Executive Director to Communications and Outreach Director to more accurately reflect her current job responsibilities.

***Family conferences.** Our fifth family conference was in Indianapolis in May 2009. We also have helped sponsor two international scientific workshops to discuss NBIA research in 2000 and 2005, and are planning a third in Oct. 2010.

***The three times yearly newsletter.** It is mailed to about 4,200 families in the United States and around the world. It is another important way for us to achieve our goal of educating others about these disorders.

***Research.** This expense covers grants, costs associated with awarding the grants, bridge funding such as the Hayflick Lab Campaign, and the BioBank program that we withdrew from in July 2009. We have funded 15 research grants totalling \$450,000 in the past eight years.

Our 2009 tax return is on our website and I invite you to review it for more in-depth information.



Hayflick Lab Campaign continues while many other fundraising initiatives underway

By Luann Rein



Luann Rein
Communications and Outreach Director

Funds are still coming in to support the Hayflick Lab Campaign, our all-out-effort to raise \$250,000 to help Dr. Susan Hayflick keep her Oregon lab open in the face of lost federal aid.

So far we have raised \$218,342 and need just \$31,658 to meet our goal. We are counting on NBIA families continuing their efforts to accomplish this before the end of this year. I am available to help you with planning your fundraiser so don't hesitate to contact me if I can be of service.

NBIA Director of Adult Programs, Mike Cohn of Minneapolis, has been working hard on several fundraising projects.

At a Dec. 31 concert fundraiser, he met Vince Corbin, a Dirt NASCAR driver who has been racing since he was 16. Corbin, 32, is brimming

with ideas to help promote NBIA disorders. For starters, he ran his first race with our NBIA logo on race car #59 on April 17. Cohn attended that race and met many of the people involved in making sure the car ran right. Little did he know that everyone working on the car was family.

Corbin's mom Cindy is also having events to help raise funds for NBIA. She organized a Texas Hold-Em tournament held on June 13 and a garage sale the same weekend, raising approximately \$500 for NBIA.



Cindy Corbin and Mike Cohn have teamed up to create some great-looking T-shirts. Many of you have seen the prototype on the NBIA listserv. Our logo is on the front left side, and on the back is a great picture of Vince Corbin and his race car. Hats will be available as well. They are white and have the NBIA logo on the front, and Corbin's signature on the back. Contact Cohn at mikecohn@yahoo.com if you would like to order either the t-shirts or hats.

Another Cohn fundraiser April 18 brought in more than \$700 for NBIA when a Minnesota organization, Community First, teamed up with The Mason Dixons and the Heartbreaker Band. Eight radio stations put Public Service Announcement's on their websites about the event thus creating more awareness about NBIA. A special thanks to the emcee Shayne Michael for helping out, and to Vince Corbin for bringing the NBIA #59 race car to the event. Alex Ligertwood, formerly of Santana, performed with the Heartbreaker Band. Cohn joined both bands on stage and sang a song with each one.

Two more fundraising events are planned in Minnesota over the next four months. The first will be the 2nd Annual Benefit Concert for NBIA at Mainstreet Bar & Grill on Aug. 7 featuring three bands, a silent auction and a live guitar auction. For more information, visit www.bandforacause.org or www.mainstreetbar.com. And on Oct. 9, the NBIA Country Music Fundraiser at The Rock in Maplewood, Minn., www.therocknightclub.com will be held.

(see fundraising on pg. 12)

Fundraising

(continued from pg.11)



Vince Corbin's race car #59 with the NBIA logo runs at Kopellah Speedway on Friday nights and Cedar Lake Speedway on Saturday nights. Both racetracks are in Wisconsin.

Vince is currently running 4th in points at both speedways in the Midwest Modified class.

In September, Cohn is planning to hold a softball fundraiser with 16 teams at an entry cost of \$150 per team. Participants can e-mail Cohn directly if interested.

The NBIA Disorders Association also is looking for new sources of funds. We are applying for a Pepsi Grant of \$250,000 at the stroke of midnight on June 30. Pepsi accepts only the first 1,000 applications each month, and it was full at 12:02 last month! When we are notified of our acceptance on July 16, yes I'm that confident, I'll let you know through the listserv, or you can look on our website at www.NBIAdisorders.org. Then you have until Aug. 1 to start the biggest "Vote for NBIA" at www.refresheverything.com campaign you can imagine!

I suggest you get familiar with the site soon, even though we're not there yet. Once the voting starts, we will need to get votes immediately. Everyone can log in every day in August to Pepsi's site and vote for our proposal, "Hope for Miracles: Support for Research to Cure Rare Disorders." We will put a widget on our website and Facebook Cause page that you can install wherever you want to remind you to vote. It's fun and easy.

E-mail your friends, family, e-mail list, church group, school group, college alums, business associates, Facebook list, sports teams you play on, other hobbyists you connect with, Twitter and anyone you think might be interested in voting for us. Remember, honor students need projects; let them find voters for us.

A special thanks to my Pepsi grant team members Wanda Fox, Sandy Leap and Megan Thomas for all their hard work on the grant, and to Alex Rennie and Bill Hartwell for their help with the video portion of the grant. Having a video is very important and significantly increases our chances of winning.

The organization is now a registered member of eBay Giving Works. If you use eBay to sell your treasures, you can now donate from 10 percent to 100 percent of the proceeds to NBIA directly by being a direct seller or a community seller. To be a direct seller for NBIA you will need to register with me at Irein@NBIAdisorders.org.

Did you know that items sell at higher prices and at a faster rate when listed with that lovely Giving Works ribbon next to them according to eBay? This is just one more way you can do what you may already be doing and contribute to NBIA.

If you held a fundraiser let me know about your events so we can share your success with everyone. Be sure to send a picture or two as well.

(see *fundraising* on pg. 13)

Fundraising

(continued from pg.12)

I recently changed my title from Executive Director to Communications and Outreach Director to more accurately reflect my job responsibilities, but everything I have been working on continues! My focus is on raising awareness for NBIA through social media such as Facebook, Twitter and other venues and supporting NBIA families and helping to raise funds for NBIA. This past year has been a wonderful learning experience and I have enjoyed getting to know everyone in the NBIA community.



Mike Cohn (right) joins Alex Ligertwood, formerly of Santana, to sing a song at NBIA fundraiser in April.



Contact Mike Cohn at mikecohn@yahoo.com if you would like to order the t-shirt here or hat pictured on pg. 11.

Both are \$15 each plus shipping



Participants at a Texas Hold-Em Tournament to benefit NBIA held on June 13 and organized by Cindy Corbin.





*Christina Lee Campbell
September 15, 1977 - March 17, 2010*

When Christina was born, we were ecstatic. We already had an amazing 4-year-old son, and we gazed with awe and wonder at our beautiful baby girl. Little did we know that the dreams we had for our family were about to change in ways we could never imagine.

Christina met the expected milestones until age 10 months, when she began having seizures. The doctors told us not to worry, but we saw the changes.

Her progress slowed dramatically. She learned to walk, feed herself and talk, although her vocabulary was very limited. She never learned to read or write, but she learned to sing and dance.

I once read that a philosopher, when asked why learning is such a struggle for some children, said something like, "Some children are better at teaching than learning." That was the case with Christina. She taught us so much more than we were ever able to teach her. She lived a life of unrestrained joy and unconditional love.

Three and a half years ago, the steady, relentless decline began. After many tests and doctor referrals, a neurologist diagnosed Christina with NBIA. The doctor told us what to expect and we were heartbroken.

During the last few years of her life, she suffered severe muscle pain and lost the ability to walk, talk, and swallow, yet she managed to smile every single day.

Her sojourn with us came to an end in March. We don't have words to describe how much we miss Christina, but we are thankful that we have 32 years of wonderful memories to treasure. And, we will always remember the many lessons she taught us about patience, acceptance, courage, laughter and love.

Elizabeth and Wayne Campbell,
and Christina's brother, Michael, his wife, Beth, and son Jonah

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

Brianna Aylesworth
Lawrence & Gloria Basista
Jennifer Bauknecht
Scott Jones
Richard & Elizabeth Thomas

Brent Bonfiglio & Family
Angela Bonfiglio Allen
Jeff Berwick
Myrian Gomez
Brigitte Rodgers

April & Tracie Flinn
Bud & Althea Brown
June McClure

Tammie Holland
Scott Holland

Michelle Isbell
Bill & Kim Patterson

Michael Komenda
Wallis Ann & Steve Strauch
Mary Komenda
James & Theresa Norbut

Brittany Leap
Francis & Kathryn Austin
Sherry Baker
Marguerite Cooper
Jeffrey & Theresa Hartzog
Judith Leap

Ashley Middendorf
Dianne Holtman
Lizzy Ludwig

Keri Patton
Paul & Janet Buhay

Jonathan Stretter
Marianne Patton
Ron & Donna Stretter

Adam Tifone
James & Sandy Jolly

Zach West
Tom Chrimer

Kimberly Wood
Jessica Crow
Gary & Barb Hampton
Arnie & Marie Mueller

In Memory Of

Bruce Belcher
Christopher Cazzato
Cheryl Lamos
Doug & Joy Lamos
Hugh Schrader

Christina Campbell
Elizabeth & Wayne Campbell
Joseph & Linda Girona
Grace Reformed Church

Wendy Devens
Dennis & Judy Devens

Madison Frederick
Bill & Patty Freeman

Brent Fry
Gene & Patricia Fry

Vicky Kelso
James & Rita Cooper

Cameron Meade
Barbara Burns-Brooks
Lawrence & Jeanne Dennin
Russell & Deanna Halsey
Donald & Dorlores Nikoden
Lisa Paige & Tina Sweet
Walter & Mary Ann Rickens

Angela & Vincent Orlando
Angela Steinker

Ben Patterson
Bill & Kim Patterson

MESSAGE FROM THE PRESIDENT



Patricia Wood

As I was looking through the articles for this newsletter, they seemed to tell a story of where we are as an organization today. My, have we grown!

There is news of our third scientific workshop/symposium to be held in October (see article on pg. 3), inviting researchers from around the world to participate. We started these with small steps and have seen our ambitions grow.

At our initial workshop in 2000, the first NBIA gene had not yet been found. but there was hope it would be soon. It was, in 2001. In 2005, at our second workshop, there were discussions on how NBIA might have common features with Parkinson's disease and there was hope in possible therapies such as Deep Brain Stimulation that had proved helpful with that disorder. Many of our families made the decision to get this surgery based on what they learned from this workshop and some have seen it help slow down the progression of NBIA.

And now, in 2010, with four genes related to NBIA already discovered and more possible, we are collaborating with another disorder, neuroacanthocytosis, or NA, to again further our knowledge in different areas.

With each workshop our knowledge of NBIA disorders has grown and this has led to new avenues to pursue. Possible treatments involving pantethine (see article on pg. 1) and deferiprone (see article on pg. 1) are being studied and give us hope for a viable treatment in the not too distant future.

Our story telling of Mark and Drew Karkourtis's visit to Washington (see pg. 8) highlights our group's increasing role in activism. We have been raising our voices in Washington starting in earnest last summer and intensifying in the past year. We are learning what is needed to get support for NBIA funding into the Appropriations Bill and to discuss our needs with our representatives in both the House and Senate.

Our new Spanish listserv (see pg. 6) that is just starting up shows how we are reaching out to another group that may not otherwise have access to support and high-quality information about NBIA treatments.

And our article highlighting fundraising activities (see pg.11) shows how far we have come in supporting research. We are closing in on our \$250,000 goal for the Hayflick Lab Campaign and we have many new and exciting ideas for fundraising for the rest of the year. Our article highlighting our 2009 finances shows how we spent the money

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.

Maria Bettinotti	Jason LaMountain
Don & Marjorie Cullen	Regina Neal
Teresa Ghianni	Jane Pickett
James Hayes	Amy Jo Rabe
Michele Isbell	Carmen & Julie Roser
Bob & Anne Knodle	Sacred Heart Church
Sandra Latham	Chris Sedlak
Steven Lee & Karen Yee	Texans Credit Union
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Hayflick Lab Campaign Donors

Lillian Cady	Quest Diagnostics
Community First	Jerri Rednour
Delta Zeta Sorority	Richard & Vicki Ruckstuhl
Goldman, Sachs & Co.	Sandra Sassaman
Michael Komenda	Andreas Savva
Rich & Sandy Leap	Demetris Terlikas
Thomas & Grova Lewis	John Venditti
Nissan North American, Inc.	Keith Verges
Peter & Carolyn O'Donnell	Karen Wing
Piedmont Country Club	

In Memory Of

Wade Patton	Ken Stromsta
Paul & Janet Buhay	Bonnie, Dick & Hardy Murfree
	AI & Pam Stromsta

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

raised (see pg. 10), and the article on Planned Giving (see pg. 9) shows how you can help us plan for the future.

Yes, all of these articles are part of a story about our progress and highlight how far we have come. I am so proud to be a part of this NBIA community and so grateful for all the support and commitment so many have shown over the years. Thank you for being part of this amazing story.



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Syndrome
Association



Our Mission:

NBIA Disorders Association is a non-profit organization

dedicated to providing emotional support to families affected by NBIA, educating the public about this disease, and monitoring and supporting research and informing others of its progress.

NBIA Disorders Association

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Visit our Web site at

www.NBIAdisorders.org

from discovery to cure

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