

Hayflick Lab Campaign Update Well on our way to our goal!

By Patricia Wood

The challenge was daunting: raise \$250,000 to keep open the lab of our premier researcher, Dr. Susan Hayflick at the Oregon Health & Science University, or see it close because of the loss of federal grants.

But here we are, eight months later, and because of your determination, the Hayflick Lab Campaign has raised \$150,690 from NBIA families and friends as of March 10, and OHSU has provided another \$50,000 in bridge funding to keep the lab going. We need just an additional \$49,310 to keep the lab open through 2011. We are hoping the federal funding climate will improve, and the Hayflick lab will receive NIH funding by that time.

Thanks to all of you who donated and who put on fundraisers in your communities. We could not have done this without you.

"I am deeply indebted to the NBIA community and the NBIADA for the remarkable efforts and outpouring of support for my laboratory research program," Hayflick said. "We are forging new collaborations and moving aggressively into new research areas that we think will lead to further federal grant funding. The NBIADA Bridge Funding is a lifesaver, which we hope to repay through new research breakthroughs to help all families with NBIA and related disorders. Thank you!"

Hayflick has committed to keeping the lab open as long as funds are available and says she now has enough funds to operate until mid 2011. She had

(see *HLC* on pg. 13)



Two research grants awarded with help from sister organization in Italy

By Patricia Wood

The NBIA Disorders Association hoped to award two research grants this year, but by year's end, the board didn't have enough money in its coffers for more than one grant. Much of the donations raised last year under the board's direction went to a successful fundraising effort to try and save the lab of Dr. Susan Hayflick in Oregon.

But then the board wondered: Could one of its two sister organizations in Europe support a second grant? Italy said yes.

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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.

Scientists from around the world to discuss NBIA and Neuroanthocytosis

Experts from around the globe will gather for two days in October to share ideas and discuss ways to collaborate on NBIA and another rare neurological disorder as a way to advance future research into both disorders.

The October 1- 2 workshop in the Washington, D.C., area is being called Brain, Blood and Iron: Joint International Symposium on Neuroanthocytosis and Neurodegeneration with Brain Iron Accumulation. It's being made possible with federal support from the National Institutes of Neurological Disorders and Stroke, the National Institute of Child Health & Human Development and the Office of Rare Diseases Research.

Neuroanthocytosis, or NA, is a group of genetic conditions marked by involuntary movements, problems with thinking and memory, progressive muscle weakness and spiked red blood cells. NA disorders are believed to be caused by a degeneration of the basal ganglia, a part of the brain that controls movement. Parkinsonism has been associated with the disorder in some patients, and PKAN, a form of NBIA, also has been linked to NA.

Dr. Ruth H. Walker of Mount Sinai School of Medicine in New York City will convene the workshop with Dr. Susan Hayflick of the Oregon Health & Science University in Portland.

The goal is to review what is currently known at the level of the cell about NA and NBIA, and from there to define NA and NBIA research priorities; determine resources that are needed to foster research; stimulate interest in research into NA and NBIA among scientists in related fields; generate collaborations between those working in related disciplines, with particular relevance to NA and NBIA; and attract early career scientists to those fields.

"The choice to partner with the NA community is intended to bring new ideas to NBIA and NA research and to stimulate new collaborations and encourage broader scientific interest in both disorders," Hayflick said. "A priority outcome of this meeting will be to develop biomedical research roadmaps for both groups of disorders. These roadmaps will identify research priorities and guide investments to enable strategic advances in NBIA and NA research."

Recent advances in NA research include the finding that a malfunction of autophagy, the body's process that removes dangerous materials from cells, may be related to brain cell death in NA and NBIA. Discussion at the Fourth International Neuroanthocytosis Symposium in the UK in July 2008 identified this as an area of potential relevance. The workshop will offer an

opportunity for experts in autophagy to study these disorders.

"I am delighted that the NINDS has determined that these rare diseases merit its support," Walker said. "This meeting provides a unique opportunity for experts in diverse fields to share information. This will also be an excellent opportunity for junior scientists to become involved in the field, as we plan to provide scholarships to facilitate their involvement."

Third annual Rare Disease Day celebrated worldwide on February 28

By Luann Rein

Attention was drawn to rare diseases, including NBIA, in a variety of observances held worldwide Feb. 28 on the third annual Rare Disease Day in the United States.

Established by the European Rare Disease Organization and first celebrated in Europe in 2008, the European organization asked the National Organization for Rare Disorders (NORD) to be its partner and sponsor for the event in the United States last year. This year it has spread to many other countries around the globe.

There are nearly 7,000 rare diseases, each of which affects fewer than 200,000 people. Nearly 30 million Americans suffer from rare diseases — one in 10 Americans.

Many rare diseases are chronically debilitating and life-threatening. Eighty percent are genetic, and 75 percent affect children. No cure exists for the vast majority of them, and many people with rare disorders face discrimination because of their disabilities, reducing their educational, professional and social opportunities.

Many go without a diagnosis or treatment, with little research or attention devoted to their disease. For 2010, the focus of Rare Disease Day was on the partnership between patients and researchers seeking to develop new diagnostics and treatments.

The NBIA Disorders Association partnered with NORD and the Children's Rare Disease Network with links to their campaigns on our Web site and FaceBook Cause pages. NBIA families have the opportunity to tell their stories and have them posted on the NORD Web site to further raise awareness about living with NBIA.

The Children's Network asked supporters to wear jeans to work the week before Rare Disease Day. Their slogan was Genes & Jeans - A Natural Fit. They also asked anyone with anything to say about

(see *rare* on pg. 4)

Rare

(continued from pg.3)

rare diseases to be part of the first ever "Blog for Rare" event.



NORD worked with Discovery Health to provide on-air and online messages about rare diseases, and activities were held in various states. A member of the Stiff Person Syndrome Association planned to sit in front on her local Wal-Mart in Oklahoma holding a poster about Rare Disease Day, while pre-medical students at the University of Connecticut planned a noontime awareness event

on their campus. A "Radio Media Tour" was to provide live radio interviews with stations across the country.

NORD also created a Rare Disease Day Research Hall of Fame and asked for nominations with letters of support. Our organization nominated Dr. Susan Hayflick and she is now a member of the 2010 Research Hall of Fame.

Against that backdrop, NBIA families and others affected by rare diseases wrote their legislators asking them not to forget rare diseases when budget time rolls around.

Grants

(continued from pg.1)

With that crucial support, the board approved two \$30,000 research grants for the 2009 grant cycle. The awards will go to Dr. Michael Kruer of the Hayflick Lab at Oregon Health & Science University in Portland, and Professor Dr. Lars Timmerman and Dr. Amande Pauls from the University of Cologne in Cologne, Germany.

The awards bring to 15 the grants our organization has funded in the past eight years. They total \$450,000, almost all of them thanks to family fundraisers and individual donations. The Associazione Italiana Sindrome Neurodegenerativo Da Accumulo Di Ferro (AISNAF) provided funding for the Kruer grant.

Italian families affected by NBIA founded AISNAF in 2006. Their president, Dr. Natale Scalise, attended our 2009 Family Conference in Indianapolis and participated in meetings with researchers there. Since then, we have had conference calls with AISNAF and our other sister organization, Hoffnungsbaum e.V. in Germany, focused on collaborating to promote NBIA research.

Hoffnungsbaum e.V. sent money in 2004 to help fund a research grant and has been a great partner. We are very happy to be working with both of these international lay advocacy organizations and plan future collaborations with them to keep NBIA research going.

Kruer's grant is titled "SNP microarray-based disease gene discovery in idiopathic neurodegeneration with brain iron accumulation." It will help find gene mutations that cause some forms of idiopathic NBIA, the name used when the genetic origin of the disorder is not yet known. Many NBIA patients do not have identifiable mutations in either the *PANK2* gene that causes pantothenate kinase-associated neurodegeneration (PKAN), or the *PLA2G6* gene, responsible for infantile neuroaxonal dystrophy (INAD) and atypical neuroaxonal dystrophy (NAD).



Dr. Michael Kruer has worked at the Hayflick Lab at OHSU since late 2008. His title is Fellow in Neurodevelopmental Pediatrics.

Idiopathic NBIA patients, nevertheless, often have disabling movement disorders, cognitive symptoms, and MRI findings consistent with NBIA. Without knowing what causes NBIA in this group of patients, it is exceedingly difficult to confirm the diagnosis, foresee upcoming challenges, or develop effective treatments. Kruer's project aims to fill this crucial gap. With improved understanding of this subtype of NBIA will come new insights into NBIA biology and treatments.

The lab will study the genes of certain families, and once researchers have narrowed their search to a critical stretch of a single chromosome where the responsible gene resides, they will perform DNA sequencing to identify it. Then, after a new gene has been identified in a single family, the researchers can sequence it in other patients with idiopathic NBIA to determine if they, too, harbor mutations. This approach has already been used to identify a new NBIA gene discovered at the OHSU lab that will be published shortly. The data suggests that several additional genes await

(see grants on pg. 5)

Grants

(continued from pg.4)

discovery. If you or your child has idiopathic NBIA and wish to be included in the search for a causative gene, please contact Dr. Kruer through Allison Gregory, M.S. at gregorya@ohsu.edu.

This approach has already been used to identify a new NBIA gene discovered at the OH SU lab that will be published shortly.

"We are extremely grateful to the NBIA Disorders Association and AISNAF for supporting this project," Kruer said. "It comes at an exciting time for NBIA research, but at a time when funding is extremely difficult to come by. With the momentum we have established already and with the generous support of the family organizations, we believe that our work will transform 'idiopathic NBIA' into recognized subtypes of the disease, a crucial step in characterizing and then developing effective treatments for NBIA."

Timmerman and Pauls will be working on a grant titled "Stimulation of the Globus pallidus internus in patients with NBIA (formerly Hallervorden-Spatz-Syndrome): prospective analysis of international therapeutic outcomes and development of a therapeutic algorithm."

This will be a prospective, open trial, multi-centre study that will look at outcomes of deep brain stimulation (DBS) in patients with NBIA. DBS is used to help alleviate severe dystonia, a painful condition common in NBIA patients. Dystonia is a term used to describe slow, involuntary, and sometimes winding movements of the limbs and trunk, occurring at rest and during action. The movements frequently are worsened by voluntary movement.

Dystonia generally is treated with oral medication, botulinum toxin and deep brain stimulation (DBS). Oral medication helps some patients, but is frequently insufficient for severe dystonia. Botulinum toxin is very good for localized, focal problems such as neck posturing or pointing of toes, but cannot easily be applied to the whole body. DBS is sometimes used when dystonia affects the whole body, and studies have shown it to be effective for other conditions causing dystonia.

In 2007 Timmerman and Pauls received a seed grant from our sister organization in Germany, Hoffnungsbaum e.V., funding a retrospective study looking at NBIA patients who underwent DBS around the world, and were able to show that DBS on average improves dystonia in NBIA patients. However, this study was limited in predicting which patients would benefit most from DBS surgery and which would not. The retrospective study was recently published in *BRAIN*, a top ranking international journal in clinical neurology. A free download of the paper is available at <http://brain.oxfordjournals.org/cgi/content/full/awq022v1>.



Professor Dr. Lars Timmerman and Dr. Amande Pauls from the University of Cologne in Cologne, Germany.

With the future prospective trial, Timmerman and Pauls hope to gain more insight into which NBIA individuals benefit from DBS. They will collect data on patients before they undergo DBS and follow them over two years. They also will collect information on the severity of the patient's dystonia, other medical problems, supportive therapy (such as physiotherapy, feeding tube, etc.), degree of disability, activities of daily life and quality of life. With the study they are looking for answers to the following questions:

- Is there an improvement in motor skills, in quality of life? Is there a reduction of disability and pain?
- Which patients benefit most from DBS? What are predictors of good therapeutic outcomes (genetic status, age at onset, patient age, course of disease, symptoms)?
- How long lasting is the effect of DBS? Is there deterioration with time?
- What supportive therapies are necessary? Can we create a treatment algorithm for DBS in NBIA patients (optimal stimulation parameters, supportive measures needed, optimal time point for surgery)?

They hope to pool data from both studies to increase their chances of being able to find predictors of therapeutic outcome. Thus, if you or your child has decided to undergo DBS as a treatment for NBIA dystonia, please contact amande.pauls@uk-koeln.de so you can take part in the study and help other patients, parents and doctors.

NBIA-Symposium in Munich gives scientists idea-sharing forum

By Angelika Klucken

A small gathering of European NBIA researchers on Nov. 28 led to an exchange of some big ideas at the Technical University (TU) of Munich, Germany.

They talked about the latest research regarding NBIA and the similarities between PKAN, a form of NBIA, and another disease, Neuroacanthocytosis (NA). NA specialist Professor Adrian Danek, a neurologist at the Ludwig-Maximilians University (LMU) in Munich, outlined the relationship between NBIA and NA. Both are neurological and are characterized by movement disorders. Scientists studying both diseases are just beginning to join forces to advance their research.

They also discussed "NBIA genes in health and disease" at the symposium arranged by Professor Thomas Meitinger, director of the Institute of Human Genetics at TU. He and his staff members, Dr. Monika Hartig and Dr. Holger Prokisch, have been working on NBIA genes for several years.



*Some participants of the NBIA-symposium in Munich:
front row (from the left): Dr. Tomasz Kmiec (Poland), Prof. Dr. Thomas Meitinger,
Dr. Monika Hartig (Germany), Prof. Dr. Ody Sibon (Netherlands),
back row (from the left): Dr. Holger Prokisch, Dr. Rüdiger Ilg, Prof. Dr. Adrian Danek,
Dr. Konrad Oexle, Dr. Arcangela Iuso, Dr. Tobias Haack (Germany)*

Professor Dr. Thomas Klopstock (LMU), Dr. Rudiger Ilg and Dr. Malte Claussen (TU). A view on neuropsychiatric features was given by Dr. Kevin Rostasy, a neuropediatrician at the University Children's Hospital in Innsbruck, Austria.

As chairwoman of the German NBIA patient organization, Hoffnungsbaum e.V., I contributed a lecture about "The Role of Patient Organizations in NBIA Research."

The meeting will be followed by a family conference in Germany in June focusing on medical care for our NBIA patients. We hope some of the same doctors and researchers will be present as they will be in October when the National Institutes of Health hosts a scientific workshop in Bethesda, Md., that will feature NBIA and NA researchers from around the world.

Overall, our scientific symposium was an inspiring exchange of information and another step forward for NBIA as it continues to gain momentum internationally.

Along with them, Professor Ody Sibon from Groningen, The Netherlands, attended as well as Dr. Tomasz Kmiec, a neuropediatrician at the Memorial Children's Health Institute in Warsaw, Poland, who takes care of a number of NBIA patients.

Lectures and discussion focused not only on clinical phenotypes of NBIA and their genotypes, but on the underlying mechanics of disease as examined in a Drosophila model of PKAN by Ody Sibon.

There also was an intense exchange about a new NBIA patient registry in Munich that will be launched soon as one part of a larger patient registry for mitochondrial diseases.

The interpretation of brain MRIs for diagnostic purposes attracted the special interest of the attending neurologists who take care of NBIA patients —

NBIA families rise to challenge of lobbying Congress for funding

By Sandy Leap

NBIA families have been hard at work for the past few months, contacting their U.S. House representatives and their senators. They have been calling, faxing, e-mailing, snail mailing and some have even had face-to-face meetings.

Many families said they received positive responses when requesting increased NBIA research funding and specific legislative language that funding for NBIA be included in the 2011 Appropriations Bill. But others said they met some opposition for seeking funds for a specific disorder. Regardless, we are getting the NBIA name out and raising awareness about the critical need for funding for rare disorders.

Congress annually considers several appropriations measures, which provide funding for numerous activities, including national defense, education, health, homeland security, and general government operations. Congress generally considers these measures under certain rules and practices referred to as the Congressional Appropriations Process. (If you are interested in learning more about the process, see <http://www.senate.gov/reference/resources/pdf/97-684.pdf>). The House and Senate versions of the bill must be cobbled into a final bill, a process that usually starts in February and ends in October.

Last year we began our awareness campaign around July. Although that was a bit late, we still made a great impact. The report language accepted into the final bill said: The Committee urges ORDR (Office of Rare Diseases Research) to put a higher priority on research involving NBIA, a disease for which there is no treatment or cure.

In December, the National Institute of Neurological Disorders and Stroke, the National Institute of Child Health & Human Development, and the Office of Rare Diseases Research agreed to sponsor a joint Scientific Workshop for NBIA Disorders and another rare disease, Neuroanthocytosis. (See article about this on pg. 3). We feel our work last year may have helped with raising awareness and getting funding for this workshop.

This year, we are being much more specific and trying to garner more support in hopes of seeing our requests make it to the final appropriations bill.

We are requesting 2.5 million for the upcoming 2011 fiscal year from the National Institutes of Health to advance understanding of the underlying cause of NBIA, generate better disease models and develop new treatments. This level of funding, as well as an additional \$2.5 million per year in fiscal years 2012 to 2015, is needed to
(see *bill* on pg. 8)

OHSU official aids NBIA In Capitol Hill efforts

By Sandy Leap



Lynne Boyle, OHSU Director of Federal Relations, from Alexandria, Va.

It may be hard to imagine, but there are people who roam the halls of Capitol Hill and get things done every day without worrying about who will get the credit.

Lynne Boyle, director of federal relations for the Oregon Health & Science University, is one of those unsung heroes. As the university's person in Washington, she is its chief advocate and bird dogs OHSU's interests, including increased federal funding for public health programs and for specific OHSU projects.

She lives in Alexandria, Va., with her family, and has also become an advocate for NBIA disorders and the Hayflick Lab Campaign. OHSU has supported the NBIA Disorders Association and Dr. Susan Hayflick's lab. Boyle was aware of the loss of federal money to the Hayflick lab and wanted to help the lab in any way she could.

But she also was touched by the day-to-day challenges NBIA families face, including my own. My daughter, Brittany, has the disorder, and Boyle took an interest in us.

"I think the real inspiration was hearing Brittany's story for the first time, as told by you and Rich last summer when we spent that afternoon on Capitol Hill," Boyle told me. "As you were describing the disorder, Brittany's symptoms and all the requirements related to her care, I was overwhelmed with emotion (OK, tears). It was because of those moments that I want to help advance the cause and research for these disorders."

Last year, she drafted report language that was included in the final budget bill for FY2010. This year she is taking it two steps further and drafting report language and funding language to help NBIA researchers receive specified funds.

Boyle also has been a wonderful teacher to me and others in the NBIA community. She is helping us understand the legislative process and how to work with members of Congress. From her, we have learned when to begin contacting the members, how to complete Appropriations Requests Forms and how the byzantine appropriations process works.

We have attended meetings with decision makers on Capitol Hill
(see *Boyle* on pg. 8)

Bill

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accelerate the pace of research and to attract new scientists to study NBIA.

Such funding would support five to ten grants of \$250,000 to \$500,000 per year for five years and would have a measurable impact on the pace of discovery and their translation to therapies.

In addition, the NBIA Disorders Association is asking that the following report language be in the 2011 appropriations bill: The Committee urges the NINDS (National Institute of Neurological Disorders and Stroke), NICHD (National Institute of Child Health & Human Development), NEI (National Eye Institute) and ORDR (Office of Rare Diseases Research) to put a higher priority on research involving NBIA, a group of rare disorders for which there is no treatment or cure. Specifically, the Committee urges the NINDS, NICHD, and NEI and ORDR to take steps to: halt the loss of NBIA researchers (including grant supplements and bridge funding); sensitize NIH grant reviewers to the public health relevance of 'limited researcher pool' disease portfolios; encourage the development of new researchers to the NBIA field; and develop a research agenda for NBIA that can serve as a guide to help drive the development of high priority scientific projects.

So far, NBIA families told me they have contacted staff of the following members of Congress:

Senate: Senator Richard Shelby, AL, Senator Blanche Lincoln, AR, Senator Mark Pryor, AR, Senator Barbara Boxer, CA, Senator Diane Feinstein, CA, Senator Daniel Inouye, HI, Senator Tom Harkin, IA, Senator Roland Burris, IL, Senator Richard Durbin, IL, Senator Mary Landrieu, IL, Senator Scott Brown, MA, Senator John Kerry, MA, Senator Al Franken, MN, Senator Thad Cochran, MS, Senator Richard Burr, NC, Senator Kay Hagan, NC, Senator Judd Gregg, NH, Senator Kristen Gillibrand, NY, Senator Jeff Merkley, OR, Senator Ron Wyden, OR, Senator Robert Casey, Jr., PA, Senator Arlen Specter, PA, Senator Jack Reed, RI, Senator Lamar Alexander, TX, Senator John Cornyn, TX, Senator Kay Bailey Hutchison, TX, Senator Mark Warner, VA, Senator Patty Murray, WA, Senator Herb Kohl, WI.

House: Rep. Jo Bonner, AL, Rep. Vic Snyder, AR, Rep. Brian Bilbray, CA, Rep. Michael Honda, CA, Rep. Duncan Hunter Jr., CA, Rep. Barbara Lee, CA, Rep. Jerry Lewis, CA, Rep. Lucille Roybal-Allard, CA, Rep. Rose DeLauro, CT, Rep. Debbie Wasserman-Schultz, FL, Rep. Jerry Costello, IL, Rep. Bill Foster, IL, Rep. Debbie Halvorson, IL, Rep. Jesse Jackson Jr., IL, Rep. Mark Kirk, IL, Rep. John Shimkus, IL, Rep. Rodney Alexander, LA, Rep. Steven Lynch, MA, Rep. Betty McCollum, MN, Rep. Dennis Rehberg, MT, Rep. Rush Holt, NJ, Rep. Nita Lowery, NY, Rep. Scott Murphy, NY, Rep. Paul Tonko, NY, Rep. Tim Ryan, OH, Rep. Tom Cole, OK, Rep. Earl Blumenauer, OR, Rep. Kurt Schrader, OR, Rep. David Wu, OR, Rep. William Coyne, PA, Rep. Patrick Kennedy, RI, Rep. Ralph Hall, TX, Rep. Gerald Connolly, VA, Rep. Jim Moran, VA, Rep. David Obey, WI.

It is still early in the process and we have a lot of work ahead. We will not know the results of our hard work until the end of the year, but we must press on and do all we can to tell our story.

If you have not had the opportunity to tell your story, or if you have a friend or family member affected by this devastating disorder, it's not too late to help. You can contact your representative or senator or contact me at rsleaper@hotmail.com to receive a template letter that you can personalize. With your help, we can find a cure.

Boyle

(continued from pg. 7)

and with the Office of Rare Diseases Research. Boyle's leadership, guidance and great big heart are reasons we have been so successful at the legislative level in such a short time.

Our organization isn't the only one that has noticed.

"Lynne has been an exemplary partner, guide, friend and advocate as we have tried to articulate the plight of families with rare disorders to our legislative leaders," Hayflick said. "She brings together a deep understanding of the ways our government works with a passion for carrying the torch on important issues. Her leadership in helping our community secure federal funding for NBIA research has been exceptional."

We will not know how the Appropriations Committee will vote until later this year, but we know we have our foot in the door because of Lynne Boyle's tireless efforts and caring heart.

New and veteran faces pitch in on end of year fundraising

By Luann Rein

The last half of the year was marked by a frenzy of fundraising activity to help keep research going at Dr. Susan Hayflick's lab at the Oregon Health & Science University. Our NBIA families rose to the occasion when Hayflick said her lab faced closure because of losses in federal grants.

The lab still needs help but is making progress. So far our organization has raised \$150,690 and OHSU has contributed \$50,000 in bridge funding, so we are only \$49,310 shy of our \$250,000 goal.

Meanwhile, here is a wrap-up from 2009 of what our families contributed in the field:

When **Stan and Norma Craig** of Milwaukie, Or., decided to throw an NBIA Benefit Square Dance in memory of their daughter

(see fundraising on pg.9)

Fundraising

(continued from pg. 8)

Donna Craig on Nov. 15, they realized their square dance family, the Bachelor N' Bachelorettes, was the perfect place to start. The club hosted the dance and had a great response from surrounding square dance clubs. All of the attendees enjoyed dancing to the calling of Les Seeley and Paula, and the cueing of Tami Helms and Tim Keck. Donna's daughter, Michelle, son-in-law, Josh, and grandson, Stone, helped with the event, including showing our new NBIA video "We Believe in Miracles."



Members of the Bachelor N' Bachelorettes Square Dance Club host fundraiser benefiting the Hayflick Lab Campaign.

The guests of honor were the research team from OHSU. Milwaukie, Or., was just the right distance for Dr. Susan Hayflick to get a square dance lesson! Dr. Penny Hogarth, Allison Gregory, Moon Yoon, and Wei-hong Xiong joined her. Dr. Hayflick's mother, sister and nieces came along for the fun.

Donna's memory was honored, and a handmade quilt was raffled. In all, \$2,217 went to the Hayflick Lab Campaign.

National Junior Honor Society member **Katie Bradbury** of Littleton, Colo., needed a community service project as a membership requirement. After talking with her parents, Sara and Tom Bradbury, Katie, 13, made a decision. Her mom knew a woman with a son who had NBIA: Cameron Meade, age 10.

On Dec. 17, Katie wrote NBIA and told us she had sent letters asking friends and family to donate money in honor of Cameron. Katie's parents had said they would match whatever she raised. She raised \$800 from just 20 people, and was able to double that to \$1,600.

Katie wrote excitedly to us about the wonderful support of family and friends, and how she hoped for a cure. And then several weeks later, another envelope came with the same handwriting. Inside was another generous check.

How many letters had Katie sent? I tried to contact Katie, but connected with her mom first.

Sara Bradbury filled in the details. She went to high school with Sally Smith Meade, but had lost touch with her during college. A mutual friend forwarded a letter from Meade asking for donations to the Hayflick Lab Campaign, prompting Bradbury to suggest the idea to her daughter. She thought the personal connection made the idea special.



Katie Bradbury of Littleton, Colo., raised \$2,200 with a letter writing campaign. in honor of Cameron Meade.

Katie wrote 50 letters to family and friends, some whom also knew Sally Meade. Her hope was that each family would donate \$10 thus bringing in \$500. And the Bradburys would match that. Ultimately, Katie more than doubled that, and so did her parents, bringing in a total of \$2,200!

The Bradburys and all of us at the NBIA Disorders Association are deeply saddened at Cameron's passing in January. We are pleased that this remarkable young boy was honored in this way.

Mike Cohn is seeing to it that many members of the NBIA family are honored throughout the year. Mike throws many fundraisers, and he came up with one we all could participate in. His idea was for the 2010 NBIA calendar you may have seen. It took lots of work and planning, and all of us sending in pictures of our NBIA family members to be featured during their birthday month.

It was such a great idea because the number of NBIA families is growing. It helps us put names to faces. This calendar was the first of what we hope will be an annual project. We enjoyed the birthday theme this year, but if you have a great idea for a new theme next year, let Mike know at mikecohn@yahoo.com. He's already looking ahead.

We ordered 260 calendars and only have 25 left. We sold them for \$10 each in the U.S. and \$12.50 outside the U.S. Norman Cohn, Mike's uncle, sponsored the entire printing of our first calendar.

Aside from the calendar, Mike is a master at incorporating his love of music into a fundraiser. As many of you know, Mike is very involved with the music community where he lives in Minneapolis. Many local bands have been helping him raise money.

On New Year's Eve, with the help of the Mason Dixon band, Mike raffled off an acoustic guitar donated by Tim Mahoney. The guitar had been signed by Mahoney as well as The Little River Band, Chris

(see fundraising on pg. 10)

Fundraising

(continued from pg. 9)



Acoustic guitar donated by musician Tim Mahoney, and signed by fellow musicians, raises \$1,000.

Brooks, Chris Hawkey, Shayne Wyatt, Shane Michael and other musicians. Mike raised \$1,000 through raffle ticket sales.

F e r g e

Frederickson, formerly of Toto and his music friends from World Classic Rockers, will be signing another guitar that Mike will either auction off or have a raffle later this year. For information on World Classic Rockers, see www.wcr.com.

On April 18, our organization is one of the many charities that will benefit financially when Medina Entertainment Center hosts the first of what we hope will be many more Community First fundraisers.

We thank Mike for his work on these projects.



Flyer for band fundraiser organized by Ben Belcher.

And speaking of using the power of music to help raise money for our cause, **Ben Belcher** while living in Japan, arranged for a concert in his home town of Rensselaer, NY to take place during his three week vacation there. "The Bruce Belcher Fundraiser Show" included performances by three heavy metal rock bands. The bands played for free at a concert organized by Ben Belcher on Dec. 19. The show was named in honor of his brother, who died of NBIA.

The proceeds from ticket sales helped the Hayflick Lab Campaign. Ben also sold t-shirts that remained from the "Bruce Belcher Bowl-a-Thon" held by his mom, Cheryl Lamos in October. Some of Ben's friends also held a bake sale in Bruce's memory. It's great to have so many people involved that honor Bruce's memory. The events raised \$320.00, in spite of inclement weather that kept many away.

A golf marathon in the fall held by Rick and Diane Tifone of Gibsonia, Pa., inspired others around them to help raise money for the Hayflick Lab Campaign.

Valerie Hengelsberg held a charity dance at her high school, Eden Christian Academy in Franklin, Pa. She was allowed to use the



Valerie Hengelsberg of Franklin, Pa., organizes school dance fundraiser.

gym for free and charged kids \$5 admission. As Valerie's Senior Project, she had to do all the planning and execution on her own. We want to applaud Valerie and cheer her on for taking the initiative to put on this very fun event and raise \$400 for the campaign.

UHL Construction, which is owned by one of Diane Tifone's cousins, was a sponsor at the golf tournament. The company also passes the hat each year for a charity at Christmas. This year the company wanted to do a

collection in honor of Adam Tifone for the campaign. The hat must have been pretty deep, because \$413 was collected that night and UHL matched it for a total of \$826.

Some of the fundraisers went beyond the U.S. borders. France, where **Valerie Bensch** and her family live, was the site of several fundraisers for her son, Romain Lerognon. A group of young students organized a game of football for the Hayflick Lab Campaign and raised \$1,100.

Valerie's factory is still continuing their efforts for the campaign and held a Sunday band concert for the whole city to attend. The concert was organized by Valerie's coworkers, and was very well attended. They are still paying expenses so we do not have a total yet, but it was considered a big success.



Friends of Romain Lerognon from Alsace, France help raise funds for the Hayflick Lab Campaign.

If you held a fundraiser in the last half of 2009 and you don't see your event here and it wasn't mentioned in the last newsletter, did I miss it? If I did, I am sorry. Make sure that you let me know about it. Be sure to send me a picture as well. I can also offer ideas if you want to hold an event. You can reach me at

Haylick Lab Campaign Donors

Thanks to all the following donors who contributed to the Haylick Lab Campaign in the past few months. We reported on most of the fundraisers listed below in the October/November 2009 newsletter and these were donations received afterward. Our past newsletters can be found at our Web site at www.NBIAdisorders.org under the literature link.

If you sent in a donation in honor or memory of someone, please check for your name on pages 12 or 14.

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Cameron Meade

September 24, 1999 - January 14, 2010

On Jan. 14, we lost our son Cameron, at the age of 10 to NBIA. Cameron had been diagnosed with PKAN two years earlier, but he never let his illness interfere with his joy of life. He left us with his amazing courage and joyful spirit.

He was an exceptional student and loved going to school at Sycamore Elementary in Holt, Mich. He attended regularly until the end of the 2008-2009 school year. Cameron's other favorites were shelling at the beach, swimming, blueberry picking, his dogs, the TV show Phineas and Ferb, and Disney Pixar films.

During the last few months, many friends, relatives, school staff, and medical personnel shared fond memories with Cameron. We also shared many memories with each other. What we have come to realize is that Cam was providing us a time for reflection and was preparing us for the realization that Life is Good is not just a slogan for a T-Shirt. What it really is, is a full recognition that true happiness comes from within and true comfort comes from sharing experiences with the people we love.

We give our most sincere thanks to Dr. Susan Hayflick, Dr. Penny Hogarth, Allison Gregory and the rest of the Research Team for their support and expertise over the past two years. Cameron was in the hospital from Oct. 16, 2009 until he came home on Jan. 6. During this time we were grateful to have the advice of Dr. Hayflick and Dr. Hogarth with regards to what care would be best for our son.

In addition, we have been supported by the staff at Sparrow Hospital in Lansing, Mich., as well as the staff at Mary Free Bed Rehabilitation Hospital in Grand Rapids. We will be forever grateful to you all.

Cameron is dearly loved and missed by his parents, Lynda and Sally Meade, sister Kylie, grandma, Aunt Susan, Uncle Steve, Uncle

Gordon, Uncle Jim and his extended family of close friends, peers, teachers, therapists and all of the loving and giving people who came into our lives because of our dear Cameron. Since Cameron's passing, friends of the family named a star after him called, "Cam the Braveheart."

"Because I knew you, I have been changed for good."
-from "For Good" from "WICKED"

In Memory of Cameron Meade

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Chloe Rae Solis

November 10, 2001 - September 2, 2009

Chloe Rae Solis, or Chlobug, as we loved to call her, was born November 10, 2001. As any parent knows, the birth of a child is the most precious gift we are given. Chloe was our little angel sent from up above.

Our journey began when Chloe was around 10 months old, and we noticed she wasn't meeting her milestones. It was not until shortly after Chloe turned 3 years old that we were informed she suffered from this very rare disease called INAD, or Infantile Neuroaxonal Dystrophy, a form of NBIA. During the seven years of Chloe's life, we were on a constant journey of physical, occupational, speech and developmental therapies, along with numerous medical appointments and procedures.

Being the parents of a special needs child brought a new perspective to our life. We were forced, but also welcomed into a very different world, filled with wonderful, thoughtful and compassionate families all struggling with "sick" children whom they loved more than life itself. It was a world we never thought we would be part of, but eventually became part of our identity and we grew to love this part of our life.

We were fortunate enough to be surrounded by wonderful therapists, teachers, aides, nurses, doctors and so many others who helped nurture Chloe along the way. We were also surrounded by countless family and friends who did their best to give Chloe the endless love that our precious little girl deserved.

Unfortunately, Chloe's life with us ended on Sept. 2, 2009, just two months short of her 8th birthday. She passed away peacefully in her sleep after a hard, long struggle with INAD.

Taking pictures has been a passion since she was born, and

therefore, we will always have a very special record of every moment of her life. At her funeral we hung hundreds of pictures of Chloe with family and friends through the years. Standing there looking at them helped us to realize that Chloe did more in her short life than most people do in a lifetime. Our family motto was to never let her disease hold us back from doing anything or going anywhere. We can say that up until the end, she lived life to the fullest, and we have no regrets.

She was our beautiful little angel with masses of beautiful brown hair and velvety soft skin. She will never be forgotten and will always remain in our hearts.

Raul and Cara Solis, along with her brothers, Reme and Rogan

Queen Bee Golf Marathon Donors

Story and additional donors in
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HLC

(continued from pg. 1)

planned to close in December 2009.

I am hoping that the families that held their first fundraiser this past year will hold another this year and that other NBIA families will join in. Even after we reach our goal for the Haylick Lab Campaign — we need funds for future research grants. We've all seen what a difference everyone working together can make.

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

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Mary Ann Roser donated at Christmas in honor of the following individuals

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MESSAGE FROM THE PRESIDENT

So many tough decisions with NBIA:
It's hard to know which way to go



Patty Wood

As always, the organization keeps me busy, but the past eight months have been a whirlwind of non-stop activity. I, and many others, have been working on the Hayflick Lab Campaign and garnering support for the federal appropriations bill so that funding for NBIA is included. We are learning how everything works in Washington and I have found it very interesting.

I took my daughter Kimberly with me when our executive director, Luann Rein, and I visited staff of each of our House and Senate members. I was encouraged by their interest and willingness to help, and surprised by Kimberly's interest in the conversations.

Usually, she complains loudly when she is bored, or she will fall asleep with the hope that something more interesting will be going on when she awakens. This time, she did neither. It felt like she was listening to the conversations, and as we discussed how NBIA affected our families, I wondered what she was thinking.

I have been thinking lately about how we are now dealing with more severe problems brought on by living with the disorder for so many years. We visited an endocrinologist who is recommending a yearly bisphosphonate infusion to help with calcium loss because of osteoporosis. Kimberly has started taking vitamin D and calcium supplements, and we recently had a body scan to check her bone density. There are some side effects to the infusion therapy and we need to weigh the benefits and risks so we can make a correct decision, I hope, soon.

Then we saw a neurosurgeon for her scoliosis, which has been getting worse. Anything over 50 degrees is considered high. She is at approximately 110 degrees.

The neurosurgeon said if Kimberly is going to live for another 5 to 10 years, God willing, a surgery to put rods in her back is necessary. It is a very big operation — and scary. I have seen other NBIA individuals quickly worsen after surgeries because of the stress it places on their body. And there is a 7 percent risk of a life-threatening infection immediately after the surgery. Her pulmonologist has warned there is a risk she might not be able to come off of the ventilator afterward.

What if this surgery shortens her life or doesn't add to the quality of it? The surgeon says it will not help with her back dystonia and the pain management doctor says the stress of surgery will no doubt make her dystonia worse. That doctor says that she should have a baclofen pump put in before the back surgery to see if it helps with the dystonia. That would require another surgery.

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.

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We would like to thank the Glen Wright Donor Advised Fund (\$5,000), and the George Fabe Fund, (\$2,000), both of the Greater Cincinnati Foundation, for their generous grants to our organization in December 2009.

Doctors also say if Kimberly's right knee is not helped by the baclofen pump, a surgery to help it bend is also necessary before the back surgery can be performed.

That means three possible surgeries in a short time frame when she has only had four in her 23 years: a hernia caused by falling on her stomach when she was two because she couldn't bend her legs to cushion the fall. A piece of glass pierced her stomach. The next was for her feeding tube when she was 3, a tracheotomy at 19, and a surgery to remove her dislocated left hip socket last year after a fracture.

I guess we have been lucky so far to have avoided more surgeries, as I know many NBIA families who have been through many more.

I read the neurosurgeon's report from our recent visit. It was strange to see names given to things I had just accepted without much thought. Clubfeet, wrist flexion contractures, right knee contracture, femur with apex anterior bowing. Right hip flexes only 45 degrees, and both elbow and wrist are held in flexed positions, with notable contractures to her fingers bilaterally. She can only hold her head rotated to the right and cannot turn it midline or to the left.

So many decisions to make, and these are not unique to my family. All NBIA families wrestle with these types of problems daily and try to keep a good quality of life while doing so.

This is why we are working so hard to fund research. No one should have to make the kind of decisions NBIA families have to make on a daily basis. Yet we must. We have no choice.



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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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