NBIA
DISORDERS ASSOCIATION
[Formerly Hallervorden-Spatz Syndrome Association]
from discovery to cure
Volume16 Issue 3
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Meetings in Netherlands, Paris show power of collaboration

By Patricia Wood

As our ties to collaborators in Europe grow stronger, research into NBIA is advancing.

That point was driven home to me twice in October when I had the opportunity to attend meetings in Europe with other NBIA advocates and scientists.

I was pleased to be counted among the 70 participants from 14 countries at the 2nd Joint International Symposium on Neuroacanthocytosis and Neurodegeneration with Brain Iron Accumulation Oct. 26 and 27 in Ede, Netherlands. Dr. Ody Sibon of

Participants at the 2nd Joint International Symposium on NA and NBIA in Ede, Netherlands.

*(see **meetings** on pg. 3)*

Pathfinder Award goes to NBIA Researcher in England

A global charity has awarded one of its first Pathfinder Awards to NBIA researchers in the United Kingdom.

The Wellcome Trust, which is dedicated to improving human and animal health, created the Pathfinder Awards to assist early-stage research involving rare and orphan diseases for which there are limited, if any, treatments.

Two Pathfinder Awards were announced in September, with the one for NBIA research totaling about £100000, which is about $158,770.

The NBIA research team includes scientists at Lilly's drug discovery center in Surrey and John Hardy, chairman of molecular biology of

*(see **award** on pg. 4)*

Inside This Issue . . .

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What is NBIA?

Neurodegeneration with Brain Iron Accumulation (NBIA) is a group of rare, inherited, neurological disorders.

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.

NBIA ALLIANCE

Worldwide Partners for a Cure

Go to: www.NBIAdisorders.org/NBIA-Alliance.htm for information and a listing of our member organizations.

Disclaimer

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

Meetings

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Groningen and Dr. Giel Bosman of Nijmegen organized the meeting to bring together researchers working on neuroacanthocytosis (NA) and NBIA.

The seeds for this exchange of ideas were planted at the first symposium in Bethesda, Md., in October 2010. Since then, we have witnessed breakthroughs in our understanding of the mechanisms underlying NA and NBIA at the molecular and cellular level. New NBIA genes have been discovered.

Two NBIA Dutch families attendng the meeting announced plans to start a non-profit in the Netherlands. That would increase the number of countries with member organizations in the Alliance to seven.

It was gratifying to meet and hear from early-career scientists who are studying these disorders. Four young researchers who produced poster presentations of research into NBIA and NA received travel fellowships to the symposium.

You might be hearing their names again. They were: Dario Brunetti from the IRCCS Foundation Neurological Institute "C Besta," Milan, Italy; Merel Adjobo-Hermans, Radboud University, Nijmegen Medical Centre, the Netherlands; Claudia Siegle-Roos, Max F. Perutz Laboratories, Medical University of Vienna in Austria and Shima Mehrabian, University Hospital "Alexandrovska" Sofia, Bulgaria.

Three members of our Scientific & Medical Advisory Board, Drs. Susan Hayflick, Penny Hogarth and Paul Kotzbauer were among the 20 scientists who addressed the group on the latest research in their fields. Ginger Irvine of the NA Advocacy group and Angelika Klucken, from the German NBIA group, Hoffnungsbaum e.V., represented the patient organizations. Irvine and her daughter told the group what it was like to live with a rare disease and Klucken discussed the patient organization's participation in the European Union TIRCON grant.

Also at the symposium, NBIA Alliance representatives discussed plans to develop a "Map of NBIA Experts" with contact details of physicians and clinicians from various countries; holding an international family conference in 2015; creating a Scientific & Medical Advisory Board for the Alliance; and developing an NBIA Health Care Notebook for families for keeping medical records and other important information about their disorder.

Two NBIA Dutch families attending the meeting announced plans to start a non-profit in the Netherlands. That would increase the number of countries with member organizations in the Alliance to seven.

NBIA Alliance representatives meet in Ede, Netherlands and welcome a new member organization from the Netherlands.
First row: Heike Jaskolka (Germany), Jean-loup Vasseur (France), Mr. Nagel-Heinen (Netherlands), Maayke van Schijndel (Netherlands), Alie Nagel (Netherlands), William Arends (Netherlands),
Second row: Antonio Lopez (Spain), Patricia Wood (US),
Angelika Klucken (Germany). Not pictured: Dr. Francesca Sofia (Italy).

Plans for a third symposium in Milan, Italy, in 2014 are now underway.

After the symposium, Klucken and I attended the Council of European Federations Workshop in Paris Oct. 30 to 31. It was organized by the European Organization for Rare Diseases, EURORDIS, which is similar to the National Organization of Rare Disorders in the United States. EURORDIS represents 537 rare disease patient organizations in 49 countries.

Klucken and I represented the NBIA Alliance, which includes all of our European NBIA organizations.

The meeting focused on the importance of patient registries in moving research forward; establishing centers of expertise for rare diseases; collaborative partnerships with other rare disease organizations to strengthen both groups; and the Rare Connect Platform, which enables organizations to create a site for promoting the organization's work. Messages posted on the site can be translated quickly by human translators into English, Spanish, German, French or Italian so that patients from different countries can more easily communicate. The NBIA Alliance plans to look into joining Rare Connect.

Research was a hot topic as well. Participants brainstormed on possible joint actions and ways to increase prospects for gaining funding from a new EU program, Horizon 2020, which is one of the main sources of research funding in Europe.

*(see **meetings** on pg. 12)*

Award

(continued from pg. 1)

neurological disease at University College London. They will develop a line of human stem cells that can be used to study NBIA, according to a news release from the Wellcome Trust.

The researchers hope the stem cell line will lead to greater understanding of what causes NBIA and accelerate the development of new treatments.

"The new Pathfinder Awards and Lilly's own collaborative approach to innovation are complementary in bringing academia and industry together," said Professor Michael Hutton, Lilly's chief scientific officer for neurodegenerative disease. "This partnership will allow us to generate research tools vital to our understanding of the functional impact of genetic mutations linked to NBIA. We are delighted to have been successful."

The other project to win a Pathfinder Award will investigate methods for delaying the progression of a rare, hereditary metabolic disorder, Homocystinuria, which leaves patients unable to metabolize the protein, methionine, which is necessary for normal metabolism.

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Long-awaited deferiprone trial about to start in Oakland

By Veronica Bonfiglio

At last, the trial to test the chelation drug, deferiprone, as a possible treatment for NBIA PKAN patients is starting.

The hematology clinic at Children's Hospital & Research Center in Oakland was abuzz with activity when my family and I visited in November. My son Brent has been on deferiprone for five years and we are firm believers in its benefits.

The clinic mailed pre-screening consent letters to the enrolled families the week of Nov. 5. With the convenience of families in mind, the clinic is making travel arrangements for families and has contracted with a company called Greenfire to provide all trial participants a credit card to use for travel expenses, such as hotel charges.

By the time families receive this newsletter, the trial should be underway. The Oakland team, led by Dr. Elliott Vichinsky, along with ApoPharma and Drs. Penny Hogarth and Susan Hayflick (via phone) were to hold a trial initiation meeting at the end of November, which is required before the study can begin.

Children's Hospital & Research Center representatives and two NBIA families participate in a meeting with neurologists about PKAN and the deferiprone trial in Huntington Beach on Nov. 1. Seated: Brent Bonfiglio, Aiden Arzate. Standing:: Kristen Giambusso, Dr. Christine Aguilar, Nancy Sweeters, Dr. Elliott Vichinsky, Veronica Bonfiglio, Gaetano Bonfiglio and Lydia Arzate.

That would clear the first families for "baseline" visits in early December. Because the team is committed to getting everyone started quickly, first visits for enrollees will continue through March.

Vichinsky's team, including Nancy Sweeters, Kristen Giambusso and Dr. Christine Aguilar, discussed the trial at a meeting Nov. 1 in Huntington Beach, Calif., to spread the word to families. Hogarth and Hayflick were plugged in by phone.

By the time families receive the newsletter, the trial should be underway.

I attended the meeting with Brent and my husband, Gaetano. We finally met Lydia Arzate and her son Ayden who is 9 and has PKAN.

Our families shared information with the group about our experiences with deferiprone. Ayden has been on the drug for 6 months, and both of our families have seen improvements.

Vichinsky included another patient, Travis Brown, and showed an ABC Eyewitness news video of Travis after taking deferiprone. It showed him making dramatic improvements.

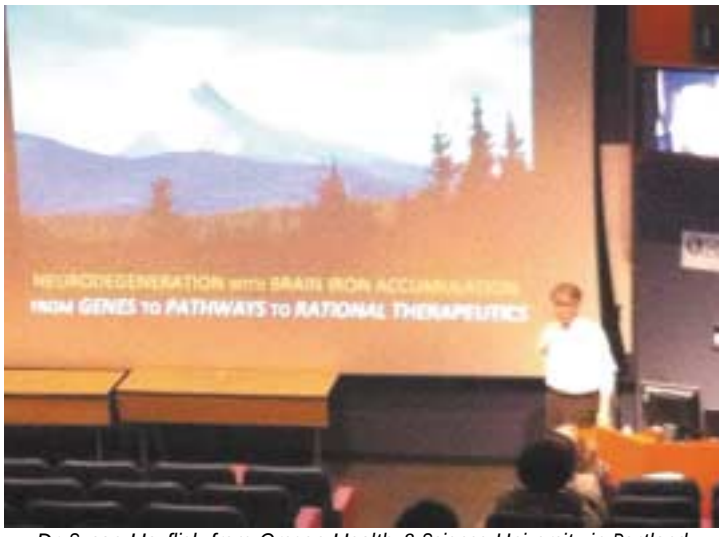
Although the number of attendees was modest, the session was well received with participating doctors asking questions of the presenters. They met Vichinsky and chatted with Ayden and Brent.

It's not too late to sign-up. Contact Sweeters at nsweeters@mail.cho.org or Vivian Ng, the PKAN- Deferiprone trial coordinator, at ving@mail.cho.org.

Premier NBIA researcher lectures, learns, in visit to top children's hospital

By Susan Laupola

NBIA's chief researcher, Dr. Susan Hayflick of the Oregon Health & Science University, visited one of the nation's top childrens' hospitals in September to listen, learn and lecture about NBIA.



Dr. Susan Hayflick from Oregon Health & Science University in Portland gives a lecture on NBIA at Cincinnati Children's Hospital Medical Center in September.

The visit to Cincinnati Children's Hospital Medical Center was the result of strategic planning by the NBIA Board late last year. The board decided it would be important for Hayflick to educate physicians and researchers about NBIA and learn what Cincinnati Children's has to offer in gene therapy resources and lessons learned about treating children with rare disorders.

The late September visit was sponsored by the hospital's Cancer and Blood Diseases Institute.

Hayflick came away highly impressed by the hospital and excited about an opportunity to partner with researchers there who specialize in producing viral packaging for gene therapy. She toured the lab where that happens, the Asceptic Processing Laboratories and Vector Production Facility, and met its director, Dr. Han Van der Loo.

"The facility run by Dr. Van der Loo is … an outstanding resource that need not be recreated elsewhere," Hayflick said. "Dr. Van der Loo was very interested in partnering with us to provide services, and Dr. (Arnold) Strauss (the University of Cincinnati's chairman of pediatrics and director of the hospital's research foundation) endorsed that idea enthusiastically. A partnership with Cincinnati Children's for gene therapy in NBIA would benefit everyone."

Hayflick was able to have a long conversation with Strauss over a dinner, which included Stella Davies, co-director of the Cancer and Blood Diseases Institute, and Dr. Joseph Palumbo, division director of hematology.

Hayflick also met with other hospital leaders, including Dr. Tracy Glauser, a professor of pediatrics and neurology; Dr. Greg Grabowski, director of the human genetics division; Dr. Jeffrey Whitsett, co-director of the Perinatal Institute; Dr. Punam Malik, leader of the molecular and gene therapy program; and Nancy Ratner, leader of the cancer biology and neural tumors program.

She had a great turnout for her lecture on "Neurodegeneration with Brain Iron Accumulation (NBIA): From Genes to Pathways To Rational Therapeutics."

"Overall, the faculty at (the hospital) were outstanding, as were the administrators," she said. "It's a very impressive place for pediatric science and health care."

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Gala puts spotlight on rare disease community

By Patricia Wood

I was privileged to attend the 1st Annual Tribute to Champions of Hope Gala held in Newport Beach, Calif., on Sept. 27 to honor those who fight every day for the rare disease community.

Not only did I enjoy the gala but I learned a lot at a patient summit held the following day, also hosted by Global Genes/RARE Project, an Orange County non-profit aimed at raising awareness and support for rare diseases.

Champion awards were given to patient advocates, scientists, doctors and biotechnology company representatives. The recipient of one of the two non-profit advocacy awards was Pat Furlong, president and CEO of Parent Project Muscular Dystrophy. Furlong was our keynote speaker at the 2009 family conference in Indianapolis where she shared her insights with our families and encouraged everyone to do their part to make a difference.

*(see **Gala** on pg. 6)*

Gala

(continued from pg. 5)

The gala inspired, educated and entertained the 400 guests. Recording artists Chris Mann and Katrina Parker from The Voice TV show performed, along with American Idol singer Elliott Yamin and singer, songwriter and patient advocate Gracie Van Brunt.

American Idol singer Elliott Yamin, songwriter and patient advocate Gracie Van Brunt, Katrina Parker and Chris Mann from The Voice TV show performed at the 1st Annual Tribute to Champions of Hope Gala in Newport Beach in September.

An estimated 7,000 rare and genetic diseases affect one in 10 Americans, about 30 million people in the U.S. and 350 million people worldwide.

The patient advocacy summit brought together patient advocacy leaders, biotech leaders in the rare disease field, representatives from the National Institutes of Health and others in the rare disease community. It also hosted a screening in Hollywood with the Kauffman Foundation to showcase a new rare disease documentary called Here. Us. Now.

The date is already set for next year's summit on Sept. 20, with the gala the following night.

This year's summit is available as a webcast. Check the website at www.globalgenesproject.org for the link.

Holiday gift certificates now available for NBIA's Path to a Cure Campaign

We all have someone on our holiday gift list that is tough to buy for or doesn't need a thing. Here's an idea: Make a donation in his or her honor to our Path to a Cure Campaign.

The campaign, with most of the money earmarked for research projects, is in the form of a virtual road trip from the Oregon Health & Science University, home of NBIA's key research lab, to San Antonio, site of our family conference in April.

You can buy a mile for $75 in honor of your loved one and receive a holiday gift certificate to give that person. Buy several miles and receive a certificate noting that a virtual key to a city, a park bench, school or other landmark has been named in their honor on the NBIA website. Details are on our website, www.nbiadisorders.org. The holiday gift certificates, which are suitable for framing, will be offered for every $75 gift made until the end of the year.

The campaign has a long way to go! Donations since August, when the campaign launched, have nudged us only about 112 miles down the road. We have another 2,555 to go by April.

We did not have our usual number of family fundraisers this fall to help this campaign along, but it's not too late to do your part. We have devised a very easy way for all NBIA families, friends and supporters to help. Simply create a fundraising page using our FirstGiving site at www.firstgiving.com/NBIAdisorders. Here's how to do it:

Put our FirstGiving site into your web browser, then click on the Path to a Cure Campaign link on the right side of the page. Next click the JOIN NOW button on the top of this page. You will need to fill out a form to create your page and once you agree to the terms and conditions, you're ready to go. You can then share the link to your page by emailing friends and family or putting a link on Facebook or Twitter so folks know that you are raising money for NBIA and need their help.

You can ask for donations in honor of your birthday, in lieu of holiday gifts, or simply post the information to help the campaign. Kristi Ose of Goshen, Ky., the mother of Jared who has INAD, created a FirstGiving fundraising page and has raised more than $2,600 for the campaign. It was very easy, she said.

"The whole process only took me about 10 minutes — to write

*(see **campaign** on pg. 7)*

Campaign

(continued from pg. 5)

his story and upload a photo. By the end of the first day I had already met 20 percent of my goal."

Ose also posted a link to the site on her Facebook page and emailed the link to friends and family not on Facebook. She pasted another link to Jared's Caringbridge site.

"Some of the people who have given are past friends of mine from college that I'm not in regular contact with. They have not even met Jared but his story made them want to give and help the cause," she said. "I feel so incredibly encouraged and blessed to see the donations pouring in."

She urged others to try, especially families who don't have time to host a 5K or some other fundraising event. "This fundraiser is the easiest and least time consuming of them all, all you have to do is post your story and hopefully you will be pleasantly surprised about what little effort on your part can bring in lots of money for such a great cause."

There are other ways you can help make this campaign a success. Fill out a PTC campaign donor form on our website to mail with a check, or make a donation by credit card at our website at www.NBIAdisorders.org.

Sponsorship Opportunities

Park Bench	**$ 300**
Key to a City	**750**
Bridge	**1,500**
City Park	**2,250**
School	**3,000**
Alien Spaceship	**3,750**
Stadium	**4,500**
Shopping Center	**4,500**

New Name Landmark: $7,500 or more
We will cross out the name on our map and replace it with yours!

Path To A Cure Donors

Miles as of 11/15/12

Mile #	
1 Gaetano & Veronica Bonfiglio	Honor of Brent Bonfiglio
2-3 Linda Erickson	Memory of Bruce Belcher
4 Anonymous	Memory of Eva Meston
5 Anonymous	Memory of Eva Meston
6 Anonymous	Honor of Brittany Leap
7-8 Kimberly Patterson	Memory of Ben Patterson; In Honor of Will Patterson
9 Anonymous	Honor of Adam Tifone
10 Kimberly Wood	Memory of George Oberg
11-12 Mary Ann Roser & Ted Thomas	Honor of Kimberly Wood
13-18 Andrew & Beverly Komenda Fundraiser	Honor of Michael Komenda
19-28 The OHSU NBIA Team	
29-32 Sue & Sam Laupola	Memory of William Arthur Staubach
33-45 Rick & Diane Tifone	Honor of Adam Tifone
46 Kyndel Ross	
47-49 Zach West Birthday Fundraiser	Honor of Zach West
50-70 Jim Carney Fundraiser	Honor of Brittany Leap
71-75 Audrey Lee	Honor of Mary Ann Roser
76-77 Mark Botten & Regina Neal	Honor of Kimberly Wood

*Ose Fundraiser miles not included as still ongoing. Current miles = 35 TOTAL MILES TO DATE: 112

Seventh family conference set for San Antonio

By Patricia Wood

We hope you'll find next year's visit to San Antonio, the Alamo City, to be unforgettable.

NBIA Disorders Association's Seventh International Family Conference will be April 4 to 7 at the Drury Plaza Hotel San Antonio North.

Once again, the family conference will include individual appointments with members of our Scientific & Medical Advisory Board. They will be held on April 3, before the conference officially starts, and April 4, with special consideration given to first time attendees. Families say they benefit greatly from this opportunity to meet with leading NBIA experts and discuss concerns about such issues as genetic testing, a sudden decline in function, significant increase in dystonia, medications, eye problems and deep brain stimulation.

Registration and family appointment information will be available by mid-December so that you can start making hotel reservations and travel decisions. Appointments are limited so they will be made on a first-come, first-served basis to registered participants.

Registration materials will be posted on our website and announced on the NBIA listserv and in e-mails to NBIA families. If we do not have your e-mail address and you would like to be notified when registration materials are available, send this information to info@NBIAdisorders.org.

For the first time, the conference will start on Thursday evening; we will conclude on Sunday at noon, as always. At happy hour on Thursday, from 5:30 to 7 p.m. at the hotel, participants can complete their registration. Afterward, from 7 to 8:30 p.m., we will have a "Getting to Know You" session that will include all families and their children. This is a great way to break the ice and meet other NBIA families. You won't want to miss this session.

If you are a NBIA adult and would like to participate in sightseeing and getting to know others a day or two before the conference starts, e-mail pwood@NBIAdisorders.org or Mike Cohn, director of adult programs, at mikecohn2226@comcast.net. Volunteers will be on hand to assist the group and act as guides.

A limited number of scholarships will be available to help defray costs for some who otherwise couldn't attend. Depending on how many apply and what funds come in, we may be able to waive registration fees and help with hotels costs, but other travel costs are not covered. Applications will be available when registration opens.

The conference will include sessions on research and services for affected individuals as well as socializing together, with a visit to Morgan's Wonderland, an amusement park for the disabled. Families always tell us how important it is to see one another again and make new friends.

We hope to have a record-breaking number of families join us in San Antonio!

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Please help with our conference scholarships

Every conference we have been able to offer scholarships to families who need help defraying their expenses. Please help us continue this tradition with a donation to our conference Scholarship Fund.

You will find a Scholarship Fund donor slip inside the contribution envelope included with this newsletter. If you prefer, you can go to our website at www.NBIAdisorders.org and use your credit card to donate, designating it for scholarships.

We appreciate all of the generous support this program has received in past years.

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Conference sponsors needed

NBIA Disorders Association always spends a large portion of its general fund putting on a family conference every other year. We keep our registration fees as low as possible so that as many NBIA families as possible can attend.

For our past conferences we have always had help from sponsors to help underwrite conference costs and we are asking once again for help. Sponsors will be listed in our conference program and will receive special recognition at the conference and in our newsletter.

Our sponsorship form can be found at our website and printed out to send in with your donation. We welcome individuals as well as businesses, and ask that you help us by sending this information to anyone you think may be interested in being a sponsor. We have different levels of sponsorship so that as many as possible who wish to help can do so.

If you have any questions regarding sponsorship, contact Patricia Wood at pwood@NBIAdisorders.org or call at (619) 588-2315.

Do you support NBIA Disorders? Take a picture and show us!

By Melissa Woods

Melissa Woods from Cincinnati, Ohio, is our new Social Media Director.

If you have been on Facebook lately you might have noticed our NBIA Disorders Association page is starting to blossom.

We are up to 300 "likes" and are continuing to expand our audience. However, we still have a long journey ahead of us before we reach 2,667 likes to match the 2,667 miles in our Path to a Cure campaign.

You can help us increase viewership on our Facebook page by participating in our new Social Media Contest. Here are the rules:

Take a photo of yourself or someone in your family holding a sign with our NBIA logo or wearing one of the NBIA T-shirts. Then, post the photo to our Facebook wall at www.facebook.com/NBIADisorders and encourage your friends to "like" your photo. The top two photos with the most likes by Jan. 31 will win a free registration — one for each winner — to the NBIA Family Conference in San Antonio April 4 to 7.

Photos uploaded before Nov. 15 will not be included in the contest. If you have already liked the NBIA page, you can still like individual photos and have them counted in the contest. Remember, the photo must include the NBIA Disorders logo somewhere.

Here is an example of Rich Leap showcasing NBIA Disorders in a Facebook photo.

Be as creative as you want with your photos. Holiday activities and family vacations are great moments to capture and can increase your likes.

If you have questions or don't know how to upload photos to Facebook, contact us at socialmedia@nbiadisorders.org . We can upload your photos for you. Happy posting!

Families raise over $38,000 at events this summer and fall

From a garage sale to an online giving campaign, NBIA families raised at least $38,613 at events they organized between July and September.

The Meade family of Diamondale, Mich., kicked off the summer/fall fundraising season by participating in a 5K run/walk July 22 at Ele's Place, in memory of Cameron Meade, who died in 2010. This non-profit provides peer support programs for grieving families and teens. Parents Lynda and Sally Meade and their daughter Kylie participated in this program after the loss of Cameron.

The Meades gave their friends "Cam's Critters" T-shirts that they wore for the event and asked that they consider making a donation to NBIA Disorders Association. They raised $725 for our organization.

The Meade family and friends from Diamondale, Mich., with their "Cam's Critters" T-shirts.

Once again, the Ose family of Goshen, Ky., turned son Jared's birthday celebration Aug. 28 into an NBIA fundraiser in his honor. They asked for donations in lieu of gifts, as they have for several years, but also created, for the first time, a personal page on our FirstGiving site to make donating online fast and easy.

So far the page at www.firstgiving.com/NBIAdisorders has received an overwhelming response, with $2,635 raised and more coming in.

*(see **fundraising** on pg. 10)*

Fundraising

(continued from pg. 1)

Kristi Ose and her son Jared, from Goshen, Ky.

"I feel so incredibly encouraged and blessed to see the donations pouring in," Kristi Ose, Jared's mother, said. "I want to encourage you all to take the few minutes it takes to set up a page. (See pg 6 for more information on setting up a FirstGiving fundraising page.)

In Jordan, Minn., Kristi Ose's friend, Rachelle Chrismer, followed in the Oses footsteps and held an NBIA fundraising birthday celebration for her son, Zach West. Zach and Jared's birthdays are only three days apart. Zach has atypical NAD, a form of NBIA.

Zach West, from Jordan, Minn., celebrates his birthday with family and friends.

Chrismer collected $280 and has since set up a personal page on FirstGiving to collect donations for our Path to a Cure campaign.

Beverly Komenda from Justice, Ill., raised $500 in honor of her son, Michael, for the Path to a Cure campaign from PartyLite candle sales this summer. Pam Stromsta from Shorewood, Ill., raised $230 for NBIA with her Avon sales. Both demonstrated creative ways to raise funds for our organization: selling goods to customers and donating the proceeds.

Peggy and Michael Wichert held a garage sale at their home in Hudson, Ohio, on Aug. 24 and 25 and were amazed by the response.

The Wichert family from Hudson, Ohio hold a garage sale to raise funds for NBIA research.
Front row: Grace Bost, Jaymes Wichert, Matt Altier
Back row: Michael Wichert, Peggy Wichert, Patty Penix, Autumn Penix.

"We thought that we would take all of the things that we could live without and put them out here (in the yard)," Michael Wichert said. None of the items for sale were priced and the family hoped that buyers would be generous and see their purchase also as a donation.

They were. The event raised $4,613.

The Fox 8 Cleveland television station did an interview with the family, resulting in donations from strangers out of state who were moved by their efforts and their son Jaymes, 22, who has NBIA. Someone posted the interview on Facebook and it became the most-watched video for the Cleveland station in late August.

*(see **fundraising** on pg. 11)*

Fundraising

(continued from pg. 10)

You can view it at http://fox8.com/2012/08/23/family-sells-everything-for-son-with-rare-disease/.

If a 100 hole golf marathon is not enough of a challenge, why not try a 100-mile run? That is exactly what Jim Carney of Front Royal, Va., did. A supporter and participant of the Leap family "Queen Bee" Golf marathon for the past six years, Carney participated in the run Sept. 8 and 9 to raise funds in honor of Brittany Leap.

The event is called the "Pine Creek Challenge," which challenged participants to see how far or how fast they could run along scenic Pine Creek in northern Pennsylvania. Carney completed the run in less than the 30 hours allotted.

"It was the hardest thing I have ever attempted," Carney said. "I want to say a huge thanks to everyone. … (and) a special thank you to my wife who paced me for the last 50 miles. I am not exaggerating when I say that I could not have finished the race without her."

Carney exceeded his $1,000 goal and raised $1,630 for our Path to a Cure campaign.

The "Queen Bee" golf tournament was held Sept. 25 after several date changes that, unfortunately, reduced the number of players but not their giving spirit.

The dedicated group of golfers enjoyed a spectacular day in the mid-70's, one of the loveliest in the event's half-dozen years. Those unable to play donated, helping to raise more than $28,000.

As always, generous sponsors and prize donors covered the full cost of the event, allowing all proceeds to go directly to NBIA. "We can't say thank you enough for those who have supported us each year," said Brittany's mom, Sandy Leap.

Jim Carney from Front Royal, Va., ran 100 miles to raise money for NBIA.

Golfers with Brittany Leap at the Queen Bee Golf Tournament on Sept. 25 in Haymarket, Virg. Front row: Mike Leap, Brittany Leap, Rich Leap, John Munies. Back row: Charlie Deliee, Chris Kase, Jay Portnoy, Tom Huard

*(see **fundraising** on pg. 12)*

Queen Bee Golf Marathon Sponsors

Marathon Sponsor

Quest Diagnostics

Hole Sponsors

Absolute Carpet Care
Bench International
Feickert & Wilkinson Family
Guenther Family
Brad Holland
Judy Leap
Gloria Lim
Teresa Markham
Mike & Linda McGraw
Cynthia & Allen Melton
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Prize/Goodie Bag Donations

My Haberdasher (Martha Cutsinger)
Panera Bread - Atlas Walk Way, Gainesville, VA
Piedmont Country Club
Quest Diagnostics - Chantilly

Sodexho (George Lee)
WineShop at Home (Maureen Colvin)
Red Fox Inn

Special thanks to Michelle Levesque of Willow Moon Healing for providing chair massages to the golfers.

Meetings

(continued from pg. 3)

Klucken presented information on the collaborative research project, TIRCON an EU funded grant that includes the NBIA Disorders Association.

I was struck by how similar US and European goals are for rare disease research. I came away with an even greater belief in our need to continue these global collaborations in our drive to cure NBIA.

Participants at the Council of European Federations Workshop in Paris Oct. 30 to 31.

Bartlomiej Janicki

January 1, 2004 - October 3, 2012

Bartlomiej Janicki, whom everyone called Bartus, arrived on the first day of 2004 to the everlasting delight of his family.

From the very beginning, Bartus was happy and smiling. His big blue eyes were always shining with pure happiness. He was cheerful and unfailingly polite, despite constant physical therapy because of NBIA.

He loved to play and have fun, but when it was time to learn and study, he was always focused He learned fast and was intelligent beyond his years.

His favorite activities were painting, drawing, building with blocks, and, of course, playing with his toy cars and trains. He did not let his illness get him down and was always willing to do anything. Even right after his deep brain stimulation surgery he was smiling and already hungry.

He loved animals, and he wanted a pet, so we gave him two aquariums filled with all kinds of fish. He found great joy in feeding them and watching them swim around freely.

To his last day, he was loved by all. He left this world in his beloved home in Opole City, Poland, his toys beside him, on Oct. 3. His life was short but very beautiful. We will always remember our exceptional child as the brightest star, our little sun.

Your loving family

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

He had a purpose and you have hope

By Pam Stromsta

Sept. 14 marked the fifth anniversary of our son's passing. Had he lived, Kenny would have been 26 on Oct. 31.

Ken Stromsta, who had PKAN, passed away five years ago this September.

Patty Wood, president of NBIA Disorders Association, recently asked me if it seemed like only yesterday that Ken was here with us. I told her, yes, at times, but it also feels like forever since we held him, or heard his mischievous laugh or saw that gorgeous smile.

I remember finding Patty through the National Organization for Rare Disorders in the early 1990's. There were only about 10 affected families then, but it meant everything to find another soul who understood what we were going through and was doing something about it.

Ken's gene mutation, which causes PKAN, was the first to be discovered by Dr. Susan Hayflick's team in 2001. That discovery was so meaningful for us.

Patty asked me to share what I would like others to know about this journey. I told her I'd try. But, I don't know if I can do Ken, or this experience, justice.

At first, it was overwhelming to learn my child had a life threatening disease and was given a prognosis of nine years. You never imagine you will have to learn to put a tummy tube in your child or care for broken legs, arrange for brain surgeries or live in a hospital for weeks. But you do it out of love, and you do it because you are given strength you never knew you had.

I believe each of us is born with a purpose. I think our children were born to show what real love, unconditional love, looks like. Ken taught me patience and to trust taking one day at a time. He showed me how to always give control of the situation to God, the only One who really knows what tomorrow will bring.

Ken taught those around him the meaning of courage and perseverance through times I can hardly bear to think about now. He taught his brother and sister that being handicapped was normal. Although I don't believe God causes a disease, He knew we

*(see **Ken** on pg. 14)*

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

In Honor Of

Brianna Aylesworth
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Ken Stromsta
Ricky & Linda Reavis
Ed & Regina Stremich
Allan & Pam Stromsta

Ken

(continued from pg. 13)

would be born into a fallen world and He turned what was meant for bad into something beautiful.

Your child will reach many people, as Ken did in his all-too-brief life. He especially touched his cousin who was teaching in a foreign country and had a terrible accident. His leg was torn apart and the pain was horrendous. He told us later, I just thought of all that Ken had been through and told myself, "If Ken can bear it, so can I." When Ken's cousin came back to the states, we let him have Ken's unused hyperbaric treatments, which he believed saved his leg.

Parents, from my heart, I want you to know, no matter how hard it was for us, it was worth every moment. You have reason for much

*(see **Ken** on pg. 15)*

Message from the President

My wish list for 2013

Patricia Wood

It's that time of year when we start thinking about gifts — what we would like to give others and what we hope to receive. It got me to consider my wishes for NBIA in the coming year.

I decided to dream big but also be realistic. We all want a cure for NBIA and that is my fondest hope. I believe we are on the right path with our researchers, but I don't expect a cure to be discovered in 2013. I do, however, wish for great progress toward that end and believe that will happen.

So, without further ado, here are my top 10 wishes (with apologies to David Letterman):

No. 10: Bring together physicians, therapists and others working with NBIA patients in a more organized way so they can learn from each other and provide better care to patients and families. I would love for our organization to sponsor training or seminars for these groups on a regular basis.

No. 9: Grow the number of countries in our NBIA Alliance. My wish is that one day we have awareness of the disorders and information to help those affected in all corners of the world.

No. 8: Have our International Patient Registry grow and include as many NBIA individuals as possible. Our goal is for the registry to lead to future clinical trials to produce treatments and, ultimately, cures for all NBIA disorders.

No. 7: Expand and strengthen all of the NBIA organizations so that we have the means to fund more research than ever before.

No. 6: Have biotech companies calling on us to work on NBIA because they know a cure is possible for many of these disorders.

No. 5: Continue the remarkable research collaborations that have evolved over the past two years and watch them grow.

No. 4: See a potential therapy for *PLA2G6* disorders be developed as researchers also advance our knowledge about all of the other forms of NBIA.

No. 3: Discover that one or more of our PKAN mouse models is just what we need to test new therapies and potential cures for NBIA.

No. 2: Find new ways of thinking about NBIA by making new connections that could be groundbreaking.

And my No. 1 wish for 2013: Find the gene that causes my daughter's form of NBIA and all the other unknown forms of NBIA. We have waited long enough!

Happy Holidays and here's to a 2013 where all of my wishes — and yours — come true!

****

Ken

(continued from pg. 14)

hope. You have knowledge and access to research we couldn't imagine 20 years ago. Use the listserv, use those of us who have been there. I would be glad to talk to anyone, especially about the things I didn't know and learned the hard way, as well as the things I did know. We still try to attend the conferences in hopes of bringing our experience and support to others.

We miss our Kenny dearly, and we will never forget the love he brought us. Sept. 14 will always be a day to remember the joy, not the pain, and be thankful that we will see our son again.

Thanks to Patty, Dr. Hayflick and all involved in NBIA for fulfilling your purpose.

" At first, something seems impossible; then it becomes improbable; but with enough conviction and support, it finally becomes inevitable."

-- Christopher Reeve

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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Donors may also be found in other sections of the newsletter under fundraisers and/or donations made in honor/memory of someone.

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Our Mission:

In our drive to find a cure for NBIA, we provide support to families, educate the public and accelerate research with collaborators from around the world.

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