

Calling all donors, participants for this year's family conference

By Patricia Wood

Registration for our Sixth International NBIA Family Conference to be held May 13 to 15 in St. Charles, Mo., is now available on our website at www.NBIAdisorders.org.

The website also has information to make your hotel reservations at the Embassy Suites St. Louis-St. Charles/Hotel & Spa; helpful travel information; and a preliminary conference program.

Families wishing to have a May 12 appointment with our doctors must fill out an appointment form. Times will be filled on a first-come, first-served basis, with priority given to first time attendees. Those seeking scholarships to help defray conference costs have until March 15 to request help by sending in the appropriate application to pwood@NBIAdisorders.org.

Donor slips for our Conference Scholarship Fund can be found 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.

Your support will help a needy family participate in this valuable experience of meeting the leading experts on NBIA, learning the latest research news and helpful treatment options and socializing with other NBIA families.

We still need conference sponsors to help us defray expenses. Our sponsorship form can also be found at our website and printed out to send in with your donation. We welcome individuals as well as businesses as sponsors at varying amounts, with recognition provided at the conference and in our newsletter.



*Embassy Suites St. Louis-St. Charles/Hotel & Spa
 in St. Charles, Mo.*

Deferiprone trial set back when U.S. denies funding

By Veronica Bonfiglio



Veronica Bonfiglio, board member from Fremont, Calif.

In what is considered a setback for families hoping to see a clinical trial of Deferiprone, a federal grant application was rejected for a trial of the iron-chelating drug submitted by a California physician.

The National Institute of Neurological Disorders and Stroke told the lead researcher, Dr. Elliott Vichinsky at the

(see deferiprone on pg. 3)

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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:

Hoffnungsbaum e.V.

Hardenberger Str. 73

42549 Velbert

Germany

Tel.: 02051/68075

Web.: www.hoffnungsbaum.de

E-Mail: hoffnungsbaum@aol.com

AISNAF

Via Fellino

5-87068 Rossano

CS Italy

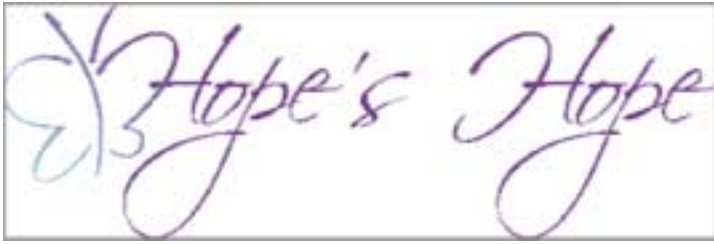
Tel: 0983.514640

Web: www.aisnaf.org

E-Mail: info@aisnaf.org

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.



Hope's Hope sponsoring conference picnic

If you are going to the family conference in St. Charles, Mo., you likely will be attending the picnic on Saturday, a huge undertaking that is the biggest social-networking event for conference participants. This year's picnic is being financed and run by one of our sister organizations, Hope's Hope (www.Hopes-Hope.org) with crucial help from the Kiwanis Club of St. Charles.

That unique pairing came about because Hope's Hope Secretary Bob Crowley also is the president of the Kiwanis Club of Apex, N.C., where Hope's Hope is based. Realizing the challenge of organizing a large picnic in Missouri from North Carolina, Crowley reached out to the Kiwanis Club of St. Charles for help in putting the event together.

Kiwanis has the goal of "changing the world one community and one child at a time" so that notion fit well with Hope's Hope mission. Hope's Hope was established in February 2005 by a dedicated group of volunteers who raise awareness and provide financial and other support to families of those with INAD, PKAN and other NBIA disorders of childhood. The inspiration for Hope's Hope was Hope Johnston of Apex, N.C., who was born with Infantile Neuroaxonal Dystrophy (INAD), a rare, genetic disorder that is part of the NBIA umbrella.

Through contributions, fundraisers, advocacy and education, Hope's Hope strives to minimize the emotional hardships and financial burdens that accompany NBIA disorders.

The menu for the picnic is still being determined, but Hope's Hope has promised an old-fashioned, all-American picnic in every way. Along with enough food to feed an army, there will be games of all types suitable to the capabilities of everyone attending. And, yes, there will be prizes for the winners.

While you're having fun at the picnic, you'll have a chance to thank the Kiwanians you will see working hard to make the event a success. You can also say hello to the team from Hope's Hope attending the conference and picnic. In addition to Crowley, President Terri Burns and Vice-President Maria Strause are planning to be there.

Deferiprone

(continued from pg. 1)

Children's Hospital & Research Center of Oakland, that it had concerns about his planned trial, including the limited number of participants, the cost (mostly for participants' travel) and that families would drop out because of the length of the trial (one year).

The institute encouraged the team to resubmit the application in July. But Vichinsky said he did not want to wait another year for a determination. So, on Feb. 2, he submitted a new grant application to the Food and Drug Administration and addressed the concerns the reviewers raised. For example, he plans to increase the number of participants from 20 or 30 patients to 40.

The FDA grants work a little bit differently than the others, and a decision is not expected before July.

Vichinsky said he is committed to helping NBIA families pursue this avenue of research and will continue to seek out every grant opportunity to make the trial happen, including resubmitting to the neurological disorders institute, if necessary.

Parents and others who are interested in advocating for the trial can contact me at veronica_bonfiglio@sbcglobal.net for the names and addresses of elected officials and decision makers at the institute and the FDA to let them know their feelings about the trial, including any concerns they may have about requiring some children to receive a placebo instead of Deferiprone in the trial.

Families, researchers, doctors gather in Italy to share knowledge about NBIA

By Dr. Natale Scalise, AISNAF president

More than 120 doctors, researchers and others, including biomedical professionals and rehabilitation therapists gathered in Milan, Italy, for a daylong seminar in February to discuss the latest research activities and leading edge therapies for NBIA.

The event featured 20 speakers and was organized by Dr. Barbara Garavaglia and Dr. Nardo Nardocci of the Foundation IRCCS, Neurology Institute "C Besta." It was jointly sponsored by "C Besta" and AISNAF, a lay advocacy NBIA organization in Italy that offers family support, NBIA education and support for research.

(see Italy on pg. 8)

Managing dystonia in NBIA patients: A treating doctor's perspective

By Dr. Tamara Zagustin



*Tamara Zagustin, M.D.
Queen Elizabeth Hospital
Charlottetown, Prince Edward Island,
Canada.*

One of the most common and disabling symptoms NBIA patients face is dystonia, a movement disorder that causes problems with walking, posture, difficulty with speech, trouble swallowing, pain, respiratory distress and sometimes spasticity and exaggerated reflexes. Because the course of NBIA is unpredictable, managing dystonia is daunting.

So far, many of the interventions that seem to improve NBIA symptoms, including dystonia, become diminished over time. Consequently, clinicians continue to work closely with families and patients to adjust treatments, with the goal of maintaining as high a quality of life as possible for patients.

The search continues for treatments that would:

- * Be successful in controlling dystonia and spasticity
- * Target the specific brain regions where these symptoms originate
- * Require the minimal effective dose
- * Provide flexibility in managing the drug or treatment, while maintaining safety
- * Allow individualization of the treatment
- * Be reversible
- * Minimize invasiveness, side effects and complications
- * Be universally available and affordable
- * Be fit for use early in life so the disease could be treated aggressively from the start

At this time, baclofen and trihexyphenidyl remain the most effective drugs for disabling dystonia and spasticity. Unfortunately, the oral forms of these medications are not very effective over time in controlling movement disorders. Moreover, side effects may crop up early on — before the drugs' full potential can be seen.

When oral baclofen is no longer effective, an intrathecal baclofen pump should be considered, probably earlier rather than later. That involves injecting the baclofen into the fluid-filled area surrounding the spinal cord. This area is called the intrathecal space. A programmable pump and catheter are surgically placed inside the body and deliver the baclofen continuously based on the settings deemed appropriate by your doctor. The pump is not without risk as infection, baclofen withdrawal or overdoses are possible complications.

On the plus side, the intrathecal baclofen pump is more effective than oral baclofen and at a much lower dosage. The treatment is reversible by removing the pump and there is flexibility in how the dosage is delivered by using a continuous or flexible mode or a combination of both modes together. The pump is less invasive than some other treatments, such as deep brain stimulation, which probably is the next best option for managing dystonia.

A baclofen pump can be used as early as age 3. It is not approved by the Food and Drug Administration for managing dystonia, but there are multiple studies and reports on its use for dystonia.

To optimize this therapy, I suggest the following:

- * The catheter tip should be placed as close as possible to the brain (high cervical region) or within the brain (intraventricular baclofen pump) although few neurosurgeons will do the IVB surgery at this time.
- * Work with an experienced team of professionals, including neurosurgeons, rehabilitation or movement disorder specialists who have experience in baclofen pump therapy.
- * Make regular visits to the doctor after the pump is first implanted, at least once every week or two, to optimize baclofen dosage within a short period of time (less than three months). Dosage should be increased in a progressive but conservative way, with increases between 20 to 30 percent on average. Usually the dose for dystonia management is much higher than the dose needed for spasticity. The upper limit is usually determined by the best response with minimal side effects.

(see dystonia on pg. 5)

Dystonia

(continued from pg. 4)

- * Implement a flex mode where boluses are programmed every three hours at relatively high dosages (usually greater than 200 mcg/bolus) with high basal rates (continuous mode).
- * Maintain good communication with the doctor in case of complications. Withdrawal is probably more frequent than overdose given the possibility of an acute pump malfunction with a baclofen dosage that is usually greater than 2,000 mcg/day.
- * At doctor visits, continue to re-evaluate the effectiveness of the treatment and the need for adjustments to optimize the therapy. NBIA's unpredictability will always be a challenge to the most experienced clinician, but before saying the baclofen pump failed in providing benefits you should explore treatment options fully.
- * Be goal-specific and goal-oriented. The goals can evolve throughout the different stages of the disease. Be realistic and recognize the limits of what can be achieved over time with the complexity of this disease. Try and be objective.
- * Other therapies such as oral medications, botulism toxin injections, and deep brain stimulation can and should be used when appropriate in conjunction with baclofen therapy to maximize outcomes and results.

More studies are needed on baclofen therapy and deep brain stimulation to improve our understanding of how and when to use these interventions for NBIA individuals, and knowing who stands to benefit the most from them. Feel free to contact me if there are any questions, comments or other at tkzagustin@hotmail.com.

Dr. Zagustin will be speaking at our family conference in May as part of our panel of doctors discussing dystonia management..

Two new members join NBIA board of trustees

By Patricia Wood

The NBIA Disorders Association Board of Trustees has two new members effective January 1, 2011, both of them parents of children with NBIA.

They are Dr. Mark Karakourtis, of Austin, Texas, and Rick Tifone, of Gibsonia, Penn. This brings to eight the number of trustees, although the bylaws allow up to 11 board members. Karakourtis and Tifone bring great experience to the board and are especially welcomed by Rich Leap, previously the board's only man.

Karakourtis is an oral and maxillofacial surgeon who has been practicing in West Austin for the past 10 years. He and his wife, Amy, have three children, the oldest of whom, Drew, age 16, was diagnosed with NBIA about five years ago.

Before moving to Austin he was in academic medicine, teaching at the University of Florida and the University of Nebraska. He received his dental degree at the University of Texas and his medical degree at the University of North Carolina.

When he's not at the office he can usually be found at home spending time with his family.

Tifone is a management consultant through his company ClearCourse Consulting, Inc. He helps business owners grow the value of their business and then plan a strategy for selling or exiting their business. Tifone has a bachelor's degree in computer science and a master's in business administration.

He grew up in Rochester, N.Y., and moved to Pittsburgh in 1980. He married his wife, Diane, in 1984. They have two children, Lauren 23, (see *board* on pg. 8)



Dr. Mark Karakourtis from Austin, Texas joins Board of Trustees. Pictured here with his wife Amy and children.

The Fundraising Beat goes on

By Luann Rein



Luann Rein
Communications and
Outreach Director

On a 40-degree Friday night in November, **Marissa Cohen** and fellow students of the **Genetics Club** at Miami University in Oxford, Ohio, set up a booth along a favorite pub crawl route and offered hot chocolate, hot dogs, cookies, donuts and information about NBIA disorders.

The students raised \$280 for the NBIA Disorders Association for their first time fundraiser and had a great time. They plan to do

another event soon for the organization and bring in even more.



The Genetics Club at Miami University in Oxford, Ohio hold a fundraiser in November to raise funds and awareness of NBIA disorders.

That same month at Bethel University in McKenzie, Tenn., **Danna Smith** and the entire **Physician Assistant Program Class of 2012** held a 5K run honoring Cole Tanner. Cole was diagnosed with NBIA in September 2009, and his mom, Johanna Tanner, is director of the PA program at Bethel.

Tanner was amazed at the outpouring of love and support that the students and co-workers have shown them since Cole's diagnosis and thanks them for all their hard work and compassion.

A total of \$1,604.83 was raised from sponsor donations, registration fees, t-shirt sales and a bake sale held during the race. Cafferty Consulting Group, LLC, Dr. J. David Outland, Cherokee Guitar Shop and Replogle Hardwood Flooring sponsored the event. Items



Danna Smith with Johanna Tanner and her son, Cole, who has NBIA.

donated to the event came from Midwest Aviation, City Florist, and Bethel University College of Professional Studies. Patrick Willis, linebacker for the San Francisco 49ers donated an autographed sports memorabilia package as a special prize.

2012 class members Kaetlin Jordan designed the t-shirts, and Emily Durrett organized the bake sale, with the sweets donated by members of the McKenzie and Paducah, Ky., communities.



Physician Assistant Program Class of 2012 at Bethel University in McKenzie, Tenn. held a 5K run to raise research funds for NBIA disorders.

Wayne and Elizabeth Campbell of Conover, North Carolina, reported strong sales of Chrissy's Candy in November and December. One local business bought 125 bags to put in their end-of-the year "thank you" baskets for their clients, while others bought candy for Christmas gifts.

The project has raised more than \$1,000 since October with help from individuals attending Life Skills, a day program for people with developmental disabilities that the Campbell's daughter Christina attended for almost 10 years. Life Skills' workers helped put labels on the candy bags.

(see fundraising on pg. 7)

Fundraising

(continued from pg. 6)



Elizabeth Campbell is selling Chrissy's Candy to raise funds for NBIA in honor of her daughter who passed away in March 2010.

Alie Nagel celebrated her 25 years on the job at Rabobank in The Netherlands by donating her entire bonus of \$942.75 to the NBIA Disorders Association. She is the mother of Jaela Nagel, who is 14 and was diagnosed with NBIA in June of 2009. Nagel earmarked her donation for research and hopes a medicine will be discovered to cure NBIA.



Jannie Heinen, Jaela Nagel and Alie Nagel from The Netherlands. Alie donated her bonus from work to the NBIA Disorders Association Research Fund in honor of Jaela who is 14 and has NBIA.

In early December, the **Brayton School Association Holiday Charity Fund** chose the NBIA Disorders Association to receive

donations, rather than holding a holiday gift swap. The organization was nominated by student Eric Brolin, who passed away later that month.

Students raised \$122 selling sweater ornaments from the school lobby Christmas tree with all proceeds going to NBIA. The sweaters were made by the students, cut out of paper and beautifully painted and decorated with bright holiday designs.

Eric had many friends at school and was proud they had chosen to do something in his honor. They included his teacher, Lorraine Brehm, and students in Room 20: Christina, Brittany, Bobby, Ben, Efrain and Dominick, as well as Eric's Room 13A teacher, Bruce Connelly, educational aide Bridget and students Kenny, Nathan, Justin, Kiwi and David.

The **Mountain View Elementary School Robotics team** in Haymarket, Va., helped raise awareness about NBIA in last year's competition on biomedical engineering. One of the areas teams were judged on was research. The Mountain View team chose NBIA, the illness affecting former student Brittany Leap.

The team e-mailed Dr. Susan Hayflick and Allison Gregory at the Oregon Health & Science University as part of its research and won second place in the regional competition. The team also sold bookmarks with information about NBIA and raised \$235.

On January 22, a snowy night in Minneapolis, Minn., **Mike Cohn** gathered friends and patrons at Toby Keith's "I Love This Bar & Grill" and raffled off a guitar. It was autographed by several 'up and coming' Nashville musicians.

The raffle raised \$900 that will go to producing the NBIA Disorders Association's compilation CD. Projected release date for the CD is April 1. Contact Mike Cohn at mikecohn2226@comcast.net for more information.

In April, Sandy Leap's friend **Heather Porter** is running the Boston Marathon in honor of Sandy's daughter, Brittany. For information on how to get Porter to the finish line, log on to <http://bostonforbrittany.blogspot.com>.

Another fundraiser in honor of Brittany will be held by **St. Raphael of Brooklyn Orthodox Christian Church**. Their annual "Queen of Hearts" Casino Night will benefit both the church and NBIA. For more information about the event, log on to www.queenbeeworld.com.

Have you held a fundraiser recently and don't see it listed here? Remember to let me know about your events so I can share your success with everyone. If you need ideas or help planning an event you can reach me at Lrein@NBIAdisorders.org.

As we continue to look for sponsors for our 2011 Family

(see *fundraising* on pg. 8)

Board

(continued from pg. 5)



Rick Tifone from Gibsonia, Penn. joins Board of Trustees.

and Adam 19. Adam was diagnosed with PKAN in 2007.

The couple's fundraising for NBIA includes a golf outing that raised over \$16,000 for research.

Tifone enjoys spending time with his family as well as golfing, skeet shooting, fishing, vegetable gardening and

wine making.

Fundraising

(continued from pg. 7)

Conference in May 2011, and donations to keep our NBIA association running, remember we have programs already in place and ready for you to start using right now. Everything can be found at our website at www.NBIADisorders.org under our "How You Can Help" link.

AISNAF

(continued from pg. 3)

It was greatly satisfying for me to welcome the unprecedented joint gathering of the scientific community and the family association.

I discussed AISNAF's activities and accomplishments, including our collaboration with NBIA Disorders Association and our participation in research funding.

Garavaglia discussed her work with diagnostic NBIA gene screening performed at the Besta Institute. Professor Sonia Levi discussed how the mechanism causing iron to accumulate in the brain is still unknown and her lab's use of NBIA patient's fibroblasts (connective tissue cells) to find answers. Dr. Valeria Tiranti reported on her team's observations of changes in the mitochondrial network of fibroblasts using a leading edge microscopy technique. Others discussed the role of ferroportin, a protein that transports iron from cells; drug treatments; and rehabilitation therapies, including deep brain stimulation and the baclofen pump.

Results of a controlled study in Italy using deferiprone on PKAN patients also were discussed, with Nardocci taking the lead and including Dr. Luisa Chiapparini and Dr. Enrico Bertini from Pediatric Hospital Bambino Gesù in Rome and Professor Alberto Piga, a hematologist in Turin.

MRIs revealed significant evidence of iron accumulation reduction in the globus pallidus in nine out of 10 patients. However, objective improvements in symptoms were observed in only one patient. This was thought to probably be due to the brief length of the six-month study.

Dr. Francesca Sofia from Telethon praised AISNAF for its work and support of research efforts. She reaffirmed her dedication and Telethon's commitment to AISNAF's initiatives. Telethon is a large non-profit organization that backs scientific research in Italy for neglected disorders. It also assists families who wish to start an organization for specific genetic disorders.

The seminar concluded with a discussion between doctors and patients about their concerns. This reinforced the necessity for a strong, constructive and complementary relationship between AISNAF and the scientific community along with the ongoing cooperation of our American and German sister organizations.



Organizers and speakers at the NBIA seminar in Italy. From left to right: Dr. Nardo Nardocci, Dr. Barbara Garavaglia, Dr. Natale Scalise, and Dr. Luisa Chiapparini.



NBIA seminar held in Milan, Italy on February 18. Discussions included the latest research activities and leading edge therapies for NBIA.



*Eric Robert Brollin
December 17, 1989 - December 21, 2010*

In memory of a favorite son and brother.

Eric was a second son with a big brother he adored. And all of us adored Eric.

He was kind, smart, fearless and determined. Those traits served him well throughout his life. He was remarkable in enduring pain and overcoming his limitations. He could communicate with expressions better than most of us communicate with speech. He had a beautiful smile and his eyes said it all. He often expressed his love with hugs, and we all looked forward to Eric's return from school — and a nice, big hug.

Eric had a wonderful sense of humor. He never missed a joke, not even the subtle ones. A slip of the tongue or a remark made without thought were enough to provoke a smile. And when his smile gave way to laughter, he pulled in everyone along with him.

He liked to 'one up' the rest of us with his exploits, and the school he attended encouraged him to be adventurous. He was the only one of us to go wind surfing, the only one to go water tubing. We've yet to see a Red Sox no-hitter, but Eric was at Fenway Park when Jon Lester pitched a perfect game in 2008. Eric even got into the dugout. He didn't have a long life, but the time he had was rich and filled with wonderful friends and caregivers.

He was always engaging, especially with the ladies. In his teens he became a decided flirt. With so many doctor appointments and hospital stays, he always found a nurse or doctor or receptionist to bat his eyes at. The elevators weren't off limits, either. They were another opportunity to engage a new friend.

It is the parents' role to teach their children, but it was Eric who

taught us patience. We drew strength from him, too. He was the brave one. He never questioned, "Why me?" He simply forged ahead with a smile, looking for fun and friends.

We are honored to have been his family and will miss him forever. We are truly grateful we were with him when he passed and that he is now pain-free and at peace. He was a remarkable boy.

Mom Cheryl, Dad Bob, Brother Kevin

Donations in Honor of Brittany Leap

- | | |
|------------------------------|-----------------------------|
| Capt. Eugene Arbogast | Brian Allen King |
| Michelle Ayala | Earle & Luzviminda Knudsen |
| Patricia Bailey | Roger & Linda Kumis |
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These donations are from various sources including fundraisers in honor of Brittany

We would like to thank the George Fabe Fund of the Greater Cincinnati Foundation for their grant award of \$3,000 on 12/9/2010 in honor of Kimberly Wood.



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

Al Aylesworth
Sandra Basista
Matthew & Nancy Cory
Mari & Kevin Johnson

Brent Bonfiglio & Family

Angela Bonfiglio Allen
Jeff Berwick

Karen Loyd

Bill & Kim Patterson

Margaret Murray

Bill & Kim Patterson

Keri Patton

Paul & Janet Buhay
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Mary Ann Roser and/or Ted Thomas

Bill Bishop & Julie Ardery
James Head
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Audrey Lee

Cole Tanner

Bethel College PA Student
Society
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Drew Trickey

Bill & Kim Patterson

Kimberly &/or Patty Wood

Allen Burnett
John & Sara Cox
Brian & Joan McMillan
Mary Ann Roser
Victoria Stapleton
Robert & Jean Starling

*Donations in honor of
Brittany Leap are on pg. 9*

Belcher/Lamos family

Patricia Paulding

April & Tracie Flinn

Bud & Althea Brown
June McClure
Jerry Oshiyama

Hannah Mercer

Robert Mercer

Jared Ose

Betty Grant
Kelly Klein
Phyllis Ose

Richards Family

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Richard & Georgette Peckham

Jonathan Stretter

James & Ellen Dick
Herbert & Anita Ford
Sarah Hunter
Monty & Gigi Jacobs
Marianne Patton
Patricia Reynolds
Ron & Donna Stretter

Adam Tifone

John & Laurretta Broderick
John & Dianne Stuckman
Richard & Diane Tifone

Zach West

Kyle Chrismer
Mr. & Mrs. Thomas Chrismer

In Honor Of

James Head donated at Christmas
in honor of the following individuals

Julie Ardery
Bill Bishop
Clare Hagerty

Virginia Hagerty
Maria Henson
Daniel LaFond

Mary Ann Roser
Ted Thomas

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

Julie Ardery
Nancy Baker
Bill Bishop
Karen Diegmüller
Deborah Duffy

Jackie Duke
Clare Hagerty
Virginia Hagerty
James Head
Maria Henson

Sharon Jayson
Audrey Lee
Lee Kelly
Kathy Monk
Katherine Tanney

In Memory Of

Sabrina & Alyssa Barbiero

Anthony Barbiero
Giulio Barbiero
Marissa Barnes
Charles Batzinger & Debbie Oriola
Michael & Linda Sciocchetti

Bruce Belcher

Linda Erickson
Doug & Joy Lamos
Carole A. Marra
Deborah Marra
Charles O'Neil

Eric Brolin

Brayton School Association
Robert & Cheryl Brolin

Christina Campbell

Lawrence & Edith Bolick
Elizabeth & Wayne Campbell

Mary & Carmen DelGrossa

Angela Steinker

Wendy Devens

Dennis & Judy Devens

Ingrid Floy

Bernard Rubin

Madison Frederick

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MESSAGE FROM THE PRESIDENT



Patricia Wood

Your NBIA board has been busy on a couple of fronts in recent months: planning the family conference in May — thanks for the strong response from those who have registered already — and collaborating on an international research grant.

Now we are ready to work on putting in place our new research agenda. You may recall that we worked with our sister organizations in Italy and Germany, along with researchers from the NBIA community since 2009, to define our research goals. In October

we announced a formal list of priorities for the next few years, and we plan to make sure those priorities are emphasized when we award the next round of research grants.

These priorities include promoting research that advances potential therapeutic uses of pantethine and deferiprone; development of induced pluripotent stem cell lines; a knock-in *PANK2* rodent model; biomarker development; and new NBIA genetic discoveries.

We hope to have the new grant application information up on our website in March for researchers to apply with letters of intent. We will then ask for full applications from those that meet our scientific criteria and will further the goals we have set.

We also plan to raise our seed grant awards from \$30,000 to \$40,000 and to consider making different award amounts for multi-year funding or special needs.

Right now we have about \$100,000 in our Research Fund. We hope to have more with fundraisers being planned throughout the year and an event in October being held simultaneously in communities in the United States and abroad. We will have more information on that at our Family Conference in a session planned by Luann Rein, our communications and outreach director.

We also will be receiving another disbursement on a \$250,000 grant received last year from a private foundation that will go toward research.

Rather than awarding the grants like we usually do in the fall, we might stagger the awards this year or hold off so we can include all funds raised in October.

Please check our website at www.NBIAdisorders.org under the Grant Info link for a detailed description of our new guidelines and a grant application. If you are a NBIA family or someone who has

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.

The Campbells thank the following people, businesses and organizations for their help and support with Chrissy's Candy sales.

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contact with a researcher who may be interested in NBIA research, please pass this information on.

With these changes, we hope to increase our pool of researchers and stimulate advances in areas of vital importance to our families and loved ones.



2082 Monaco Ct.
El Cajon, CA 92019-4235

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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

2082 Monaco Ct.

El Cajon, CA 92019-4235

E-Mail: info@NBIAdisorders.org

phone: (619) 588-2315 fax: (619) 588-4093

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