NBIA
DISORDERS ASSOCIATION
[Formerly Hallervorden-Spatz Syndrome Association]
from discovery to cure
Volume18/Issue1
February/March2014

Board awards 3 research grants totaling $239,680 to study NBIA

By Patricia Wood

The NBIA Disorders Association board is awarding $239,680 to three research projects that received top scores from a pool of 22 grant applications.

After receiving recommendations from grant reviewers with the organization's Scientific & Medical Advisory Board, the trustees selected projects that scored highly for scientific merit and are expected to lead to greater understanding of four distinct forms of NBIA.

The grants will further research into PLA2G6-Associated Neurodegeneration, an NBIA disorder known as PLAN, Mitochondrial-membrane Protein-Associated Neurodegeneration, or MPAN, Beta-propeller Protein-Associated Neurodegeneration, or BPAN, and a novel, new NBIA gene.

In addition to these grants, the board plans to make a special round of awards to study the most common form of NBIA, Pantothenate Kinase-Associated Neurodegeneration, or PKAN.

Drs. Joanne Ng, Ahad Rahim, Manju Kurian and Esther Meyer from the University College London in London, England, are researchers working on two NBIA grants recently awarded by our organization.

A call for applications will be made after the trustees conclude a strategic planning session in late April and confers with our Scientific & Medical Advisory Board on goals for the PKAN studies.

Here are details of the three awards:

Our largest-ever grant, $150,000 for a three-year study titled "Novel Therapeutic Strategies in NBIA: A Gene Therapy Approach for PLA2G6-Associated Neurodegeneration (PLAN)," could make a life-changing difference in the lives of those living with PLAN. The award goes to Dr. Manju Kurian, working with Drs. Ahad Rahim, Joanne Ng and Simon Waddington from University College London and Great

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

Together for Tomorrow far exceeds campaign goal

We are grateful to everyone who helped make our recent Together for Tomorrow NBIA Community Campaign a roaring success.

TOGETHER FOR TOMORROW
AN NBIA COMMUNITY CAMPAIGN

The campaign raised $243,000, far more than the $135,000 goal we had set. Support came from over 340 individuals and also included

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

Inside This Issue . . .

SCIENTIFIC & MEDICAL ADVISORY BOARD

Officers/Board of Trustees

Board

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 a 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/family-support/NBIA-Alliance for information and 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.

Grants

(continued from pg. 1)

Ormond Street Hospital in London, England.

PLAN includes infantile neuroaxonal dystrophy, atypical neuroaxonal dystrophy and adult-onset dystonia-parkinsonism. "There are currently no disease-modifying treatments for our patients with PLA2G6-related disease, and developing a new treatment strategy is an absolute priority," Kurian said.

Her team will use the PLA2G6 mouse model, which is an excellent disease model of PLAN, to develop a gene therapy strategy aiming at curing or significantly improving symptoms in the diseased mice. The team has expertise in gene therapy and will use a "viral vector" as a delivery system to replace the faulty gene with a working gene. That way, the normal PLA2G6 protein is restored to the parts of the body that need it: the brain and the nerve cells. The researchers hope this strategy paves the way for future clinical trials in patients with this form of NBIA.

A requisite for a clinical trial in gene therapy is a detailed understanding of the disease, so the researchers will also undertake natural history studies to understand the clinical course and long term outcomes of those with PLAN.

The research group is part of a wider team at University College London (UCL), which has a track record in bringing such "proof-of-concept" laboratory studies to clinical practice.

A second grant, valued at $44,680 titled "Functional Characterization of Newly Identified NBIA Disorders Using Neuronal Cell Models" also will go to Kurian. She will be working with Dr. Esther Meyer, also at UCL. They plan to study how genetic mutations cause disease in patients with BPAN.

They also will study the genetic mutations that cause disease in a new NBIA subtype that is the topic of a soon-to-be-published gene discovery.

In this project, they will convert patient skin cells into neurons (brain cells) and study these cells. The ultimate aim is to test new drug compounds in the cell model to see if they improve the signs of disease, which, if successful, could lead to the development of new therapies for patients with these NBIA conditions.

Finally, a $45,000 grant is going to Dr. Ana Messias, working with Drs. Arie Geerlof and Arcangela Iuso from the Helmholtz Center Munich, Germany, for the project titled "Three-dimensional structure determination and preliminary function analysis of C19orf12, a protein involved in NBIA."

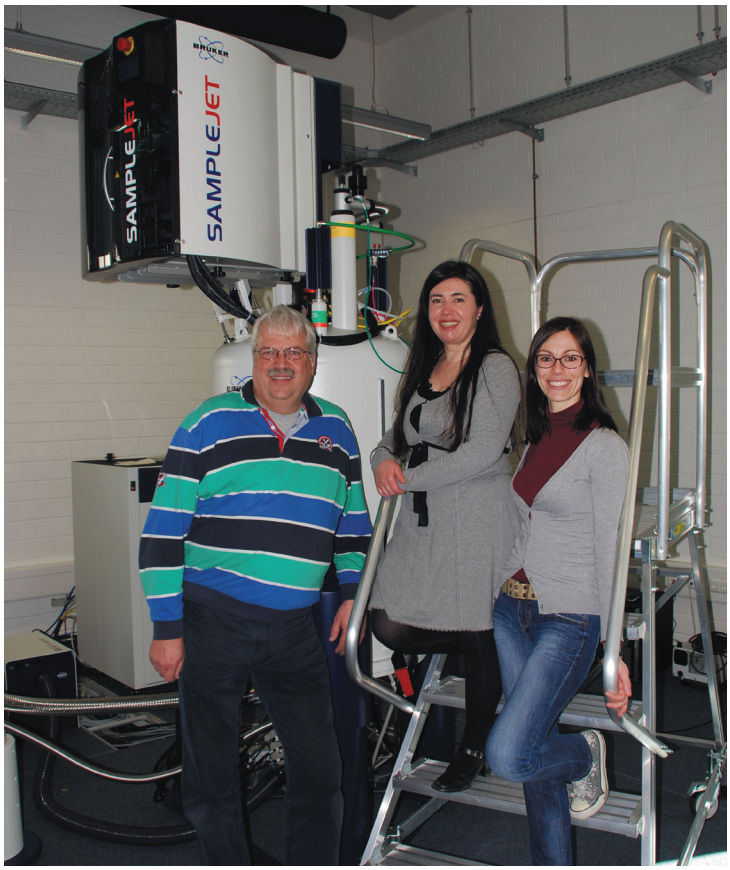
Messias and Geerlof are structural biology experts and Iuso is part of the team that in 2011 identified the MPAN gene.

Since the identification of the *C19orf12* gene, several mutations have been identified as being responsible for MPAN, but how that happens remains a mystery, as the gene has no known similarities with other known genes and its function in the cell is unknown.

This research proposes to unravel the function of the *C19orf12* gene by studying the structure of the protein it encodes. They will do this using Nuclear Magnetic Resonance (NMR) spectroscopy, a method based on the same principles as an MRI, which is used in medicine for imaging the human body. X-ray crystallography will also aid in determining the structure of the protein. This method is centered on the diffraction patterns produced by X-rays upon passing through protein crystals, a phenomenon also used to produce holograms on credit cards.

It is hoped that the information gained will allow the researchers to unravel what the C19orf12 protein does in the body using software analysis, which compares it with other known proteins with known function. They will also do biochemical testing of possible functions of the protein in the laboratory to better understand its function.

The goal is to see how mutations lead to disease and, ultimately, uncover possible therapies for patients with MPAN.



Drs. Arie Geerlof, Ana Messias, and Arcangela Iuso from the Helmholtz Center Munich, Germany, received a $45,000 grant to study MPAN. Behind them is the NMR spectroscopy machine that they will use in their research.

Campaign

(continued from pg. 1)

a very generous anonymous gift and a corporate donation from our friends at Retrophin, a partner in our NBIA research and a sponsor at our family conference in San Antonio.

Money from the campaign has helped the board to fund three substantial research grants and to issue a call for a second round of grants this year. See article on page 1.

"We are thrilled with the success of the campaign and grateful for the generosity of the NBIA community," NBIADA Board President Patricia Wood said. "We would like to thank all the donors and volunteers who stepped up to the plate and generously supported this campaign. It's amazing what we can accomplish if we work together."

Development Director Amanda Hope encouraged individuals and NBIA families to set up personal fundraising pages through FirstGiving. Those pages helped make the campaign so successful.

A first time NBIA fundraiser, Kristin Phillips, not only raised over $10,000 including matching gifts in just a few days using FirstGiving, she was also instrumental in securing a $150,000 donation from an anonymous donor.

"Also instrumental to the success of the campaign was a matching donation of $25,000 from Retrophin," Hope said. "Many people learned about the matching grant and were excited about the opportunity to double the impact of their donations."

Several individuals and families also held fundraising events, dedicating the money raised to the campaign.

NBIADA board members Rick Tifone and Rich Leap collaborated on a golf tournament that brought in $33,000.

Mike Cohn, front, with Shayne Michael, DJ Kid George, and DJ Bubba Rocks at fundraiser in Saint Paul, Minn. in February for our campaign.

Mike Cohn organized a music-related event at the Hoggsbreath Restaurant and Bar in Saint Paul, Minn. Cohn tied his FirstGiving page to a drawing at the Hoggsbreath for a guitar signed by over two dozen country music artists. The guitar was donated by the Jacob Martin Band.

With help from social media volunteers Melissa Woods and Jennifer Cleary, Cohn used Twitter and Facebook to reach out to a broad audience of country music fans. The result was an impressive $6,250 in donations. Winning the guitar was Dr. Horacio Plotkin, chief medical officer at Retrophin. He plans to use the guitar to raise awareness for NBIA by displaying it at the company's New York City office.

Tanner Young celebrated his 7th birthday with a party and donations earmarked for the campaign. In addition, donations from a fund called Tanner's Troopers set up by his parents, Scott & Niclole Young, contributed to the campaign and together they brought in over $5,300.

Sue Laupola, vice president of the NBIA Disorders Association, said she appreciated each individual donor as well as the employers who contributed matching funds. "The NBIA community was very supportive this year, and we appreciate their donations and the confidence they have in us," she said.

For a list of campaign donors since our last newsletter, please see page 5.

FirstGiving Campaign Fundraisers

Ritzman Family - $11,255
Phillips Family - $10,990
Mike Cohn - $6,250
Kimberly Wood - $2,255
Davis Family - $1,220
Susan Laupola - $1,200
Judy Pike Smith - $670
Tina and Lyle Eidelbes - $650
Jarmila Trnka - $455
Melissa Woods - $50

Tanner Young, center, from Cheswick, Penn., celebrates his birthday and raises funds for campaign.

Together for Tomorrow Campaign Donors

Anonymous (51)
Mary Beth Abramson
Pam Adam
Lisa Aguirre
Kristin & Dan Anderson
Linda Baer
Ken Banta
Anthony Barbiero
Marek Biestek
Big Wheel Brigade
Stephen & Kathy Billiot
Patsy Bolduc & Family
Linda Bosse
Bradow Family
Burkhalter Family
Aidan Byrnes
Deborah Calvert
Wayne & Elizabeth Campbell
Don & Carrol Carter
Erik Caspersen
Mary Ann Chazan
Jason Chew
Chimera Research Group
Elisabeth Christensen
Jonathan Cohn
Lourdes Conkright
Sandy Conlon
Mary Ann Cropper
Jen Crownover & Family
Dolly & Bob Dahl
Rick & Deann Dahl
Ross & Beth Darden
Steve & Robin Dawson
Harold & Carolyn DeLaMare
Mary DeLaMare-Schaefer
Kristen Demarco
Gin & Daniel De-Mesa
Jimmy Donaghy & Family
Melissa Ebig
Tiina & Lyle Eidelbes
Mary & Rod Eidelbes
Mike Eidelbes & Family
Cruz & Nora Esquivel
Jacob Evans
Kenny & Carolyn Faires
Faires Plumbing Co., Inc.
Tom Fernandez
Ryan Filotei
Brian Fix
Dan Franzese
Stephanie Freda
Mike Fuentes & Family
Galen & Terry Gaither
Robert & Sheila Galbreath
Nancy Gaughan
Al Geller
Mark Giebel
Vladimir & Michelle Golovine
Serge & Aleksandra Golovine
Goodworth Family
Dhesha Govender
Fabiana Grant
Jim & Joyce Gray
Evan Greebel
Maddie Grimsley
Margie Hattox
Jackie Hausfeld
Lawrence Hawkins
Harriet & Jeff Hayes
Robert Henderson
Jimmy Hengel
Rhonda Hinman
Natalie Holmes
Shayna Horwitz
Nicholas & Ty Jitkoff
Tatiana Jitkoff
Dave & Ann Johnson
Betty Johnson
Kathy Kelchner
Hyo Tan Knebel
Paul Koffy
Nancy Kotzian
Kristi Kratsa
Al & Lorene Kreusel
Katie Laupola
Max Laupola
Raymond & Shanna Lee
Little Bear Investments
Liu Family
Dorothy & Greg Lurie
Troy Mali
Emilio & Denise Martinez
McKesson Foundation
Bill & Diane Meehan
Kimberly Mitchell
Lori Mitchell
Gina Moore
Diane & Charlie Murdock
Ken Naylor
Rebecca Ne
Regina Neal & Mark Botten
Carolyn Nelson
Pam Nelson Harte
Joan Oetken
Kiki Ovcar
Troy Ovcar
Jong , Linda & Mitchell Park
Virginia Parker
Alison Persichetti
Marc & Barbara Pestien
Kay Peterson
Shelly Pfleger
Wylie Phillips
Andrew & Kristin Phillips
Susan & Tom Phillips
Dasha & Wylie Phillips
Martha & Bob Pike
Horatio Plotkin
Jeff Pixel
Hans Plate
Horacio Plotkin
Sheryl Poole
Zachary Prensky
Judy Pike Smith
Quail Run Stables
Karen Reese
Jay Rehurek Family
Steven Richardson
Julie Ritzman & Family
Carol Rocheleau
Erin Rocheleau & Family
Liz & Miranda Roland
Mary Ann Roser
Kyndel Ross
Katie Rossi
April & Charlie Sanchez
Chris & Kali Schmidt
Jiri Sefranek
Bobby Shea
Martin Shkreli
Jimmy Silverman
Larry & Peggy Slate
Sandra Isabel Smith
Sara Smith
Victoria Stapleton
Joani & Ed Staubach
Vera Stehlikova
Christie Stevenson
Adela Stockova
Adrienne Stuckey
Ed Sullivan
Tanner's Troopers
Holly & Rob Tennent
Mimi Test
Christi Traver
Pam Traver
Jarmila Trnka
Rafaela Typaldos
Andrew Vaino
Connie & Joe Voss
Kathy Vuturo
Tricia Waldrum
Lori Walseth
Amanda Wear
Peggy Wichert
John & Bobbie Wirth
John & Amber Wirth
Melissa Woods
Michele & Art Zamora
Mark & Cindy Zollar

NBIA Fundraisers

The Bethel University Physician Assistant Program in McKenzie, Tenn., continued its tradition of support for NBIA research, hosting its fourth annual NBIA 5K in October. The run had a Halloween theme and was part of a fall festival, with a costume contest and activities for kids. The event raised $2,300 in honor of Cole Tanner, who has NBIA and whose mother was the former program director at the university.

A group of Quality Assurance team workers at Intersections Inc. in Chantilly, Va., raised $700 for our organization. Jyoti Gupta, mother of Sahil Goyal who has PKAN, is part of the team and the effort to raise funds.

Tanner Young on left with friends. An October Disney-themed party and luncheon held by Tanner Young's family and friends in Cheswick, Penn., raised $2,913 for the NBIA Disorders Association.

We have many differences but are more alike than not

By Veronica Bonfiglio

Veronica Bonfiglio, board member from Fremont, Calif.

The various NBIA Disorders are like an alphabet soup — PKAN, MPAN, BPAN, PLAN, FAHN — each with their own distinguishing characteristics, but there's a common link: all affected individuals have elevated iron in the basal ganglia and axonal spheroids.

I don't come from a scientific background, and sometimes I have to wonder whether there are more differences or commonalities among the different forms of NBIA. I have concluded that we are all pieces of the same puzzle, making it essential that we see the big picture and NBIA's various forms as pieces of the whole.

Let me explain, with help from the National Institutes of Health's Genetic Home Reference website. If a gene's instructions for making a protein are changed, a mutation can cause the protein to malfunction or to be missing entirely. When a mutation alters a protein that plays a critical role in the body, it can disrupt normal development and lead to illness and genetic disorders.

Even though the gene mutations are different, at present researchers think that people affected by any form of NBIA may end up on a common biological pathway.

Wikipedia explains that a biological pathway is a series of actions among molecules in a cell that leads to a certain product or a change in a cell. Such a pathway can trigger the assembly of new molecules, such as a fat or protein. Pathways can also turn genes on and off. Some of the most common biological pathways are involved in metabolism, the regulation of gene expression and the transmission of signals.

Allison Gregory, a genetic counselor with Dr. Susan Hayflicks's Lab at the Oregon Health and Science University, gave an example of shared biologic pathways in our NBIA disorders.

We are all part of the same puzzle. If one piece is missing, we can't solve it... Our board firmly believes that a breakthrough for all could come from studying any one of the identified forms, including those where the gene mutation has not yet been identified and are classified as Idiopathic NBIA.

The protein in the *PANK2* gene, which causes the PKAN form of NBIA, localizes to the mitochondria and is involved in the Coenzyme A pathway and lipid, or fat, metabolism. The gene *C19orf12*, which causes MPAN, codes for a protein that also localizes to the mitochondria, but we don't yet know what the function of this gene's protein is. The protein made by the gene *PLA2G6*, which causes PLAN, is also involved in lipid metabolism. Clearly, there are themes related to the function of the mitochondria and lipid metabolism. The problem is that we still don't know "how they all hang together," Gregory said.

Gregory says that the discoveries of *PANK2*, *C19orf12*, *PLA2G6*, *WDR45* (BPAN), and *FA2H* (FAHN) help investigators when trying to identify other potential NBIA genes which may also be involved in mitochondrial function or lipid metabolism.

Gregory described the current state of knowledge about NBIA disorders as "having a handful of puzzle pieces but missing the majority. The ones we have don't even hook together; we're missing the ones that go in between."

As a mom and NBIA board member, I have no doubt that we are all part of the same puzzle. If one piece is missing, we can't solve it. As such, each piece is equally important to the others. For this reason, our board firmly believes that a breakthrough for all could come from studying any one of the identified forms, including those where the gene mutation has not yet been identified and are classified as Idiopathic NBIA.

That approach is good for research and good for our families who may experience isolation, stress and limited resources. We are stronger as individuals and as a group when we work together.

We have been able to develop international connections that have resulted in the creation of sister organizations in seven countries and the formation of the NBIA Alliance that advocates worldwide for all forms of NBIA. The European Union's TIRCON research grant shows the power of collaboration between industry, research institutions and our patient advocacy organizations. The shared goal is to improve the lives of all those living with NBIA disorders.

Both our Scientific & Medical Advisory Board and the Board of Trustees are more convinced than ever: We can and will solve this puzzle together.

NBIA research advances with Hogarth grant award

Dr. Penny Hogarth from OHSU recieves funds for NBIA research from the Oregon Clinical & Translational Research Institute.

NBIA researcher Dr. Penny Hogarth has been awarded startup funds to pursue a new area of NBIA study and to develop tests for gauging severity of the illness.

Her group received the awards from the Oregon Clinical & Translational Research Institute for developing projects that demonstrate exceptional clinical and translational promise. Translational research involves taking basic science discovery gained through laboratory experiments and "translating" that information into useful medical practices.

Hogarth's NBIA Research Group at the Oregon Health & Science University will develop and test scales for rating the impact of NBIA on affected individuals to improve understanding of how NBIA symptoms progress.

The team also will investigate an intriguing connection between a newly discovered form of NBIA, Beta-propeller Protein-Associated Neurodegeneration (BPAN), and Rett syndrome, a neurodevelopmental disorder that affects girls almost exclusively. Those affected have normal early growth and development followed by a slowing of development, unusual hand movements, slowed brain and head growth, walking problems, seizures and intellectual disability.

To develop and test rating scales specific to NBIA, the group will use a set of tools recently developed by the NIH. These tools measure motor, sensory, cognitive and behavioral function. In other words, how people move, feel, think and behave. The tests are designed to be short and simple.

Using this system, individuals being studied and their families can access the online rating scales from the comfort of their homes.

For example, subjects could be asked "Are you able to go for a walk of at least 15 minutes?" The individual would then pick the answer that best describes his or her situation: without any difficulty, with a little difficulty, with some difficulty, with much difficulty or unable to do.

Having the ability to rate the natural progression of NBIA is critical to future NBIA-related clinical trials. This information will help researchers and physicians understand if therapeutics administered during a clinical trial are helping.

BPAN shares certain features with Rett syndrome, including seizures, loss of language skills, unusual hand movements and impaired sleep, as well as some genetic features. It is hypothesized that some individuals that meet the clinical criteria for Rett syndrome, but lack mutations in the Rett gene *MECP2*, will be found to carry mutations in the BPAN gene *WDR45*.

The NBIA Research Group at OHSU is working closely with the Rett syndrome community in the U.S. and Europe in hopes of identifying these individuals.

****

NBIA group's development director steps down after 15 months of service

After 15 months of service with the NBIA Disorders Association, Amanda Hope of Gibsonia, Pa., has resigned as the organization's first development director for family reasons, effective April 25.

Hope worked with the board to create a fundraising strategy and was primarily responsible for researching funding opportunities, submitting grant applications to corporate and private foundations and maintaining relationships with key donors. She was instrumental in transitioning the organization's donor data to a secure online donor management software program, which will enhance efficiency as well as security.

She spearheaded the association's recently completed successful community campaign, Together for Tomorrow, which raised over $243,000 in funds for NBIA research and support services.

"I am so grateful for the opportunity to have worked for such an amazing organization," Hope said. "I have been privileged to meet many wonderful, dedicated, and inspirational people over the past year. It is with great regret that I have to resign from my position as development director, but I am looking forward to working alongside the NBIA community as a volunteer for many years to come."

The board greatly appreciates Hope's many contributions and looks forward to a continued relationship with her. Please direct any fundraising or volunteer questions to info@NBIAdisorders.org.

In Memory Of

Sabrina & Alyssa Barbiero
Sciocchetti Family

Eric Brolin
Bill & Bridget Horan

Christina Campbell
Wayne & Elizabeth Campbell
James & Rosalie Raley

Donna Craig
Drs. Susan Hayflick & Penny Hogarth

Wendy Devens
Judy & Dennis Devens

Sharon Haugen
Kathy Kelchner

Cameron Meade
Jann Nestell

Arleen Meyer & Ken Stromsta
Mr. & Mrs. James Brussette
Alan & Sherry Butterfield
George & Bonnie Dvorak
Mr. & Mrs. Emmett Gray
David & Jason Greyer
William Greyer
Gerald & Paulette Hartding
Mr. & Mrs. Jim Kudelis
Natalie & Eric LaStrong
Mr. & Mrs. Dickenson Murfree
Mr. & Mrs. Rick Reavis
David & Alice Sylvester

Aeva Marie Wzest
Anonymous (2)
Central Engineering
Christine Creamer
Continuum Pediatric Nursing Services
Sharen K Duff
Ryan Filotei
Kube Tech Custom Molding, Inc.
Steve Wirrig

Becky & Bruce Belcher
Oakley, Linda & Ashleigh Bathricks
Arthur & Myrna Carkner
Donna & Christopher Cazzato
Deborah Hirschklau
Doug & Joy Lamos
Charles & Karen O'Neil
Sharon Oppedisano
Anna & Joseph Riley
St. Peter's Hosp. Ambulatory Surgery Unit
St. Peter's PACU Staff
Hugh Schrader

Carmen & Mary Del Grossa
Angela Steinker

Robb Eidelbes
Tina & Lyle Eidelbes

Lisa Higgins
Shelly Allen

Wade Patton
Paul Buhay
Cyndy & Mark Patton

Bernie & Richie Roberts
Judy Roberts

Julie Smyth
Maxine Smyth

Syd Walsh
John Walsh

Kathy Annette Wells
Victor & Karen Wells

Zach West
Anonymous
Alger Family
Jessica Arbour
Dana Rae Barr
Sarah Bauer
Wendy Berry
Bowring Family
Burke Family
Bernadette Chrismer
Rachele Chrismer
Karla Cramer & Family
Dyrhaug Family
Amanda Franklin & Family
Heather Fry
Becky Geis & Family

In Memory Of

Zach West (cont.)
Bill & Mary Glosemeyer
Debra Haefner
Pat Halvorson
Lisa Hauser & Family
Jen Hedrington & Family
Heyer Family
Leah Holmberg & Family
Neil Jackson & Family
Lynea Kalenze & Family
Anita Laffen
Nicole Langheim & Family
Angie Lederle
Alex & MaryBeth LeFevre
Spencer Lindholm
Matt Lorenz & Family
Jacob & Laura Menden
Katie Micklo
Vicki Miles
Sandy Morgan
Amanda Ohmes & Family
Carrie Pena
Sharon Pierce
Nancy Rudd
Laurna Schultz-Schmidtke
Janae Skare & Family
Rick Svihel
Tracy Swanson
Kennedy Thompson
John & Becky Walker
Chrissy Walker
Amy Westerlund & Family
Tim Young & Family
Geri-Anne Zubich

Sarah L. Wronko
Anonymous (5)
Karen & Bruce Adelman
Jackie Bacilo
Diane Bartlow
Billheimer & Raquet
B. Theodore & Helen Bozonelis
Michael & Margaret Brown
Diana Cangemi
Dennis & Vincene Caporusso
Kate & Patrick Chauvin
Alfred & Jeanann Consoli
Judy Crystal
Greg & Joan Damell
James & Sandra Darcy
Mary Ann & John Daudt, Jr.
Robert & Kathryn DeFillippo
Theresa & Gerald Delguercio
Richard & Margaret Dieckhoff
Education Assn of Mt. Olive
Lawrence Fox
Mr. & Mrs. Robert Gaynor
Barbara & Eugene Geller
Terry Haynes
Bob & Ellen Heigl
Mark & Gail Holleran
Kenneth & Barbara Hyman
Katy & Matthew Jablonowski
Johnson & Johnson
Edward & Joanne Kays
The Kerri Jones Family
Sanford & Sonia Kaplan
Noreen I. Kelly
Cindy Kenworthy
Alexander & Patricia Lobban
Glenn & Dawn Maguire
Julia Margolin
John & Sarah McGrath
Mt. Olive Child Care & Learning Center
Mt. View School Sunshine Club
Peter & Karen Olivieri
David & Rosa Pipher
Irina Polotskiy
Re/Max Heritage Properties
William & Mary Reynolds
Ridge Family
Nicole Ritchie
Charles & Ada Roth
Rix Pool & Spa
Erin Ruffo & Carol Stark
St. Clares Hospital (CM & SW Depts)
Sandshore School
Kathleen & Robert Sanger
Regina & Robert Sanger
Anne Shakespeare
Sam & Christine Simone
Jennifer & Mark Slater
Arthur & Maryann Smith
John & Rita Toohey
University of Scranton
University of Scranton Graduate Assistants
Janice VanHorne
Michael Vavra
Audrey & Michael Wallock
Betsy Werthan
Andrea Wesolowski
Rosemarie Wilkinson
Andrew Wubbenhorst
Joseph & Diana Zanko

Rebekah (Becky) L. Belcher
May 20, 1977 - November 12, 2013

Becky was a very happy child who grew up to become a loving, caring adult. She was a loyal friend and a wonderful sister.

Becky was my oldest child and helped me take care of her siblings as best she could. I will miss her beautiful blue eyes and dazzling smile.

She loved to read and gather trivia on The Beatles. When she lost the ability to walk and talk, I enjoyed reading to her. I also would play and sing Beatles songs to her. She often responded with smiles and happy noises.

Throughout her life, Becky had a marvelous faith in the Lord. I know she is now walking on streets of gold in heaven. She was an angel waiting for wings. Now, she has them.

Love,
Mom

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

Kathleen Annette Wells
October 31, 2013

Sarah L. Wronko
March 18, 2014

Aeva Marie Wzest
February 26, 2014

Eva Dögg Ólafsdóttir
September 19, 1990 - February 20, 2014

When Eva was born in September 1990, it was like the whole world got brighter. When she smiled, which was often, it felt like the sun was knocking softly on the window.

But these last few days all of us who knew her have been shrouded in darkness. Even the sky seems to be gray in the sunshine.

Eva passed away in her sleep on Feb. 20. We know the grief will slowly fade — not because we stop mourning or thinking about her, but because, bit-by-bit, the joy will take over. It is the joy of having known her, the joy of all the beautiful memories we will always cherish.

During her short life, she suffered more pain than most of us will ever know, but her serenity and her smile was there until the end.

A few hours after Eva died, her closest relatives and friends gathered at her bedside. Some were crying, and some of us might have been a little angry at the higher powers. The priest came, we all joined hands, and the circle closed with Eva's mother holding her hand and the priest putting his hand on Eva´s forehead. This moment convinced me that no soul has ever been accompanied by more positive thoughts.

I know that Eva, where ever she is, is now running and singing her favorite tunes. She is eternally surrounded by love and affection, like she was all her life.

With love from your parents and grandparents,

Harpa Dís Harðardóttir, mother Ólafur F. Leifsson, father
Hugrún Skarphéðinsdóttir, grandmother
Hörður Björgvinsson, grandfather

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
Alfred & Laura Aylesworth
Sandra Basista
Debbie Erickson
Linda & Bryan Kohles

Brent Bonfiglio
Sharon Amon
Jeff Berwick
Angela Bonfiglio Allen
Gaetano & Veronica Bonfiglio
Julie Cerna
Paula Dauget & Family
Terri Foster
Praveen & Reeta Gupta
Kuldeep Kalonia
Ali & Parvaneh Kheymehdooz
Nancy Lueder Misra
Mr. & Mrs. Gene Mochizuki
Piotti Family
Zumcorp Inc.

Renee Jamros
Raymond Jamros

Brittany Leap
Lisa Aguirre
Susan Cooper
Samuel & Linda George
Deborah Heritage
Judith Leap
Janet Nolle
Quest Diagnostics
Vivian Votipka
Colleen Wood

Jared Ose
Phyllis Ose
Kandi Tahmoush

Libby Rattray
Kenneth Shaw

Brandon Richards
Chuck & Beth Morgan
Della Reasbeck

Board of Trustees
Drs. Susan Hayflick & Penny Hogarth

Christina Brant
Vincent Brant

Paul Burkhardsmeier
Wayne & Nyoka Chandler
Michelle Warden

April & Tracie Flinn
Joyce Judd

Devaj Goyal
Vineela Guntuku

Sahil Goyal
Vineela Guntuku
Jyoti Gupta
Gauri Pathak
QA Team Members
Mary Punitha Sekar

Drew Karakourtis
Dean & Lori Crutchfield
James & Melinda Davis
Don & Lena Gibson
Dr. Michael & Orla Karakourtis
Jean Karakourtis
Entrepreneurs Foundation
James & Lynelle Richardson
David Zodikoff

Sonja Olson
Rob & Sara Olson

Keri Patton
Paul Buhay
Cyndy & Mark Patton

Mary Ann Roser
Anonymous
Julie Ardery & Bill Bishop
Nancy Baker
Jacqueline Beasley
Maria Henson
Lee Kelly
Robert Schultz

In Honor Of

Mark & Martha Nell Richards
Roy & Vicki Deskin
Georgette Peckham

Kennedy Thompson
Gilbert Breaux

Adam Tifone
Tony Gallagher
David & Kathleen McRandal

Jaymes Wichert
Curtiss & Penny Tschantz

Tanner Young
Chris & Laurie Coffman
George & Rene Demko
Sandra Drakicki Bell
John & Sophia Facaros
Peter & Colleen Katz
Patrick & Lisa Reilly
Tanner's Troopers

Martin Shkreli
James Silverman

Jonathan Stretter
Herb & Anita Ford
Jacqueline McPheeters
Marianne Patton
Ron & Donna Stretter

Madeline Verges
Anonymous

Kimberly & Patty Wood
Anonymous
Allen Barnett
Margie Hattox
Lori Mitchell
April Penera
Gary & Carol Steinker

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

Julie Ardery
Nancy Baker
Bill Bishop
Karen Diegmueller
Deborah Duffy
Jackie Duke
Clare Hagerty
Virginia Hagerty
James Head
Maria Henson
Sharon Herman
Audrey Lee
Kathy Monk
Katherine Tanney
Patty Wood

Mark & Martha Nell Richards donated at Christmas in honor of the following individuals

Mr. & Mrs. Stacy Cogbill
Mr. & Mrs. Charles Morgan
Dr. Cordell Klein
Dr. & Mrs. Richard Peckham
Luke Richards

NBIA families are reminded of the need for brain tissue donation

It can be a hard subject to discuss, but our families are reminded that their loved ones can continue to help find a cure for NBIA even after death.

In the quest to better understand what causes NBIA, researchers need the opportunity to study the brains of affected individuals. Being able to access such critically important information is instrumental, not only to better understanding of the disease but to finding better treatments.

We hope you will consider such a donation to the University of

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

Message from the President

Patricia Wood

I've been following the many leads and dead ends in the search for the missing Malaysia Airlines plane for more than a month. Hope rises that an answer is at hand, only to be dashed soon after. This has happened repeatedly over the course of the investigation and theories and facts seem to change daily.

The families waiting for news of their loved ones anguish over the uncertainty and look for answers. How did this happen? Why, who or what is to blame?

How does this relate to NBIA? It reminds me of the search by many families for answers, for some way to help the loved one in their lives who has NBIA. They must be like the plane investigators, turning over every clue in their search.

An investigator's function is to obtain information and evidence. This is something NBIA families do continuously, especially those with children who cannot voice their needs or their pain. We must be sleuths who follow a trail looking at how everything fits together to solve a puzzle. We examine each piece, systematically, in an attempt to learn the facts about something that is not obvious, not even to the doctors and specialists we turn to for help.

Our family has experienced this twice in the last six months. First my daughter was having breathing problems in November, suddenly requiring supplemental oxygen for no apparent reason. We did all the usual tests and followed the trail to find the culprit. It took six weeks of diligent effort to figure out that her carbon dioxide levels were extremely high, a problem we solved with a Bilevel positive airway pressure, or BiPap machine.

Again in April, we made the rounds of doctor visits and tests. Sudden severe bouts of dystonia made us think the baclofen pump my daughter, Kimberly, has to control spasms was not working correctly. Then, a urinary tract infection was the suspect, followed by a kidney stone. More tests followed and more negative results.

Doctors placed a catheter because Kimberly's bladder wasn't working correctly and we didn't know if that was caused by the anesthesia during a recent surgery to change her pump battery or the pain medication she was prescribed afterward. Or, perhaps neither of those was the cause.

We got no certain answers and could only do what parents in our situation would do. We investigated every lead in the hope that one will lead to a solution. Kimberly's catheter was removed after a week, and the severe dystonia has ceased. So, maybe that was the culprit, though we were told it doesn't usually cause severe pain. And maybe the dystonia will start again and we will be back to the drawing board.

We stopped the pain medication in case that was the cause of the bladder trouble, as it was listed as a possible side effect. Her bladder seems to be working somewhat, and we are hoping it continues to improve.

Yes, NBIA families have earned the title of investigators, and we do it out of necessity. It is one of our many jobs as we take care of our loved ones to the best of our ability. Sometimes we solve the mystery, and sometimes we never do.

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.

Anonymous
Bethel Univ. Phys. Asst. Program, Class of 2015
Jaroslava & Maria Bochkovich
L.J. Brehl
Joseph & Inez Crawford
Burtchaell Foundation, Inc.
James & Ellen Dick
Gartner Matching Gift
Dennis Gort
Richard & Sally Graham
Frances Klosterman
John & Vera McMahon
Carlos Medina
Mary Nixdorf
Amy Pace
Lisa Paulson
Pepsico Foundation Employee Giving Programs
Quest Diagnostics Matching Gifts
Jane Pickett
Jillian Reed
Anella Roser
Carmen & Julie Roser
Elaine Simms
Ray & Ardell Skoglund
Robert & Jean Starling
Michael Trombley
Robert & Sandra Wintringer

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

A special thank you to the Glen Wright Donor Advised Fund of the Greater Cincinnati Foundation for their grant award of $5,000 on 12/12/13 and the George Fabe Fund of the Greater Cincinnati Foundation for their grant award of $3,000 on 12/27/13.

Brain

(continued from pg. 10)

Maryland Brain and Tissue Bank. NBIA researchers have made significant advances such as identifying the presence of abnormal protein deposits in the neurons of those affected by PKAN. This discovery would not have been possible without this precious tissue.

If you are willing to consider such a donation, you can fill out a form online now and not have to worry about it later. These forms can be found at http://medschool.umaryland.edu/btbank/family/.

****

NBIA
DISORDERS ASSOCIATION
2082 Monaco Ct.
El Cajon, CA 92019-4235

Return Service Requested

Formerly Hallervorden-Spatz Syndrome Association

NBIA
DISORDERS ASSOCIATION

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.

NBIA Disorders Association

2082 Monaco Ct.

El Cajon, CA 92019-4235

E-Mail: info@NBIAdisorders.org

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

Visit our Web site at

www.NBIAdisorders.org

from discovery to cure

Formerly Hallervorden-Spatz Syndrome Association