



New Board Members Gerry Barbiero of Niskayna, NY, Phil West of Vancouver, Wash., and Treasurer, Jeff Doerner of San Diego, Calif.

NBIA Disorders Association has three new board members

By Patricia Wood

Three men have joined the formerly all-female NBIA Disorders Association board, bringing a variety of business expertise and personal experience to the organization.

They will replace Treasurer Gayle McMahon of El Cajon, Calif., Secretary Debbie Forstall of Los Angeles; and board member Dianne Gray of Naples, Fla. Each has completed five-year terms on the board. We are grateful for their service.

Board member Mary Tapke of Cincinnati has taken over the duties of secretary, and our new treasurer—and one of our new board members—is Jeff Doerner of San Diego.

Jeff is a retired pharmaceutical industry executive who has served on various other boards as president, vice president and treasurer. He works part time for a CPA firm doing accounting and tax return preparation.

Jeff has a bachelor's degree in biology with an emphasis in biochemistry. He also has a master's of business administration with an emphasis in operations management. He brings expertise to the board in medical science as well as business and accounting.

In his free time, Jeff enjoys reading, bicycling, researching his family genealogy and volunteering with charitable organizations.

A second new board member Gennaro "Gerry" Barbiero is the father of two girls with NBIA, Alyssa, 8, and Sabrina, 9, who passed

(see Board on pg 5)

Fundraising has slowed dramatically; research grants for 2005 in doubt

By Kris McGourthy

We had fun runs, bowling tournaments and raffles that raised more than \$200,000 in three years, enough to finance six research grants and cover the cost of administering them. We deserve a collective pat on the back. But 2004 has been a much slower year for fundraising. And the prospects for research grants for next year look doubtful.

We all live incredibly demanding, busy lives. Most of us care for at least one disabled child, and some of us have two or more children affected by NBIA. We are pulled in many different directions, and frustration with the disorder is high. Most of us would give anything for a cure, but we feel powerless.

But there is one thing we all can do to fight back. We can raise money to help the doctors and scientists working on this disease get closer to finding a cure for NBIA. Many of us have already shown what a little creativity, hard work and a little help from our friends and family can do. We just need to get more of you to do your part.

Even if you don't have a family member affected by this disorder, you can help in some small way. Reach out to your church; ask if the youth group would be willing to sponsor a car wash or bake sale for NBIA. Tell them, for some of our children, it could make all the difference.

You may even find someone wanting to help with a bigger fundraiser like a bowl-a-thon, 5k run or a family fun day, Imedia about your event. Newspapers and TV stations are looking for feature events on the weekends when news is slow.

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Disclaimer

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

Southwest Airlines ships 'elevator' chair to NBIA family clear across the country

By Nancy Overman

When a friend of mine in Northern California gave me an electric chair for going up and down the stairs, it was the perfect thing for my niece's NBIA-affected children in Massachusetts. But getting the gift to them was a challenge I could not have imagined.

I could not find anyone who would ship the 300-pound chair to the East Coast at a price I could afford. In desperation, I called Southwest Airlines. There, I found Rich Smith, the head of cargo for Southwest in Sacramento. He told me that since Sept. 11, the airlines no longer ships items for private citizens. I told him I was special. Rich was special, too. He listened to my story.

I told Rich my son-in-law, Clifton Von Buck, worked for Southwest, and this lift was going to a good cause. After I told him about Michael and Bobbie McGourthy and how they could use the lift to get up and down the stairs, he went way beyond the call of duty.

Rich spent two weeks working on my problem and would not take no for an answer. All I know is that somehow, he and his boss, Mike Hafner, director of ground operations in Sacramento, twisted arms and worked a miracle.

When Rich called me and told me how to have the chair wrapped for shipping, I cried. Southwest sent it to Providence, R.I., absolutely free. My sister picked it up from there. Rich is my hero.

In his honor, and in honor of the McGourthy family, I am sending a gift to NBIA's Research Fund. I can't think of a better way to say thanks.



Rich Smith, head of cargo at Southwest Airlines in Sacramento

Patients and doctors now have two options for testing *PANK2* defect in NBIA cases

By Allison Gregory



Allison Gregory

The Oregon Health & Science University DNA Diagnostic Lab launched a new test for NBIA-affected individuals with pantothenate kinase-associated neurodegeneration (PKAN) in June. Doctors and families will now have two options when considering where to have patients tested in the U.S. to see if they have the PKAN form of the disorder.

They can now choose to have the testing done at the University of Chicago, where it has been done since 2002, or at OHSU.

Dr. Susan Hayflick and her colleagues at OHSU and the University of California at San Francisco discovered the PKAN gene in 2001. Because Dr. Hayflick runs a research laboratory that does not perform diagnostic testing for patients, she has never been able to formally offer testing to the many families with which she has worked. Until now.

Through a collaboration with Dr. Sue Richards, the director of the DNA Diagnostic Lab, this testing will now be available at OHSU. Richards has extensive experience in molecular genetic testing and recently joined OHSU after serving as director of the DNA Diagnostic Lab at Baylor College of Medicine in Houston.

The PKAN test uses only a small blood sample. Blood can be shipped to the lab from anywhere in the U.S. and often from other countries. Once the lab receives it, the DNA is removed from the blood cells and amplified so that the lab has a larger amount to study. The lab then screens the PKAN gene for mutations, or typos, in its code.

After this initial screening, the entire gene is sequenced; meaning each letter of the code is checked individually. Unfortunately, few DNA tests are able to find all the possible mutations in a gene. However, the PKAN DNA test will find 98 percent of mutations in people who have the eye-of-the-tiger sign. This is the tell-tale magnetic resonance imaging (MRI) change that we expect to see in people with PKAN. The combination of an MRI and DNA testing is the most reliable way to diagnose this rare condition.

The results are available in two to three weeks and are reported to the doctor who orders the test. In addition to testing blood, the lab can test cells from an amniocentesis or chorionic villus sampling to provide prenatal diagnosis during a pregnancy. The test

costs \$1,080, and in many cases, the lab is able to bill private insurance or Medicaid. Individuals should contact the lab and their insurance company to explore coverage.

The Oregon lab can be reached toll-free at (888) 375-4636; the University of Chicago lab can be reached toll-free at (888) 824-3637.

Update on Research in Chicago

A Chicago researcher who received a grant from NORD, sponsored by the NBIA Disorders Association, reports that he is making progress in using a mouse model to study a form of the disease known as pantothenate kinase-associated neurodegeneration, (PKAN) which affects about half of the people with NBIA.

Dr. Han-Xiang Deng of the Northwestern University Feinberg School of Medicine is using gene-targeting techniques in his research. The goal of his project, for which he was awarded a grant, is to learn how chemical changes contribute to the disease so that useful therapies can be developed.

Deng said his lab's first two attempts at gene-targeting techniques did not succeed, but that the work yielded useful information. "We have made considerable progress in targeting the *PANK2* gene responsible for PKAN," he said. "Our current strategies are based on the lessons learned and we should soon have the knock-out mouse model for PKAN."

If the work proceeds as planned, the new mouse model will enable Deng and his crew to study how PKAN develops and "a rational approach to its treatment," Deng said.

Fundraising

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Even if you can't do a fundraiser, don't be afraid to contact your local media to tell them about yourself and your child. Or, if you're an adult with the disorder, you also have a story to tell and you can raise awareness about NBIA in the process.

When you tell your story to the media, explain how we are trying to raise money for new research and for our next family conference and scientific workshop in 2005 where researchers from around the world will be gathering to discuss future research on our disease. Let the media know how important it is to be with other families because of the support you gain.

I am convinced that fundraising is something we can all do, and you will be amazed at the sense of empowerment you will feel by doing something to advance the fight against NBIA. It is vital that we keep the research going. Please help by doing your part.



Sabrina Barbiero

September 11, 1994 - March 21, 2004

The birth of our precious daughter, Sabrina, was one of the greatest moments of our lives. This March we lost her to NBIA. By the time she died at age 9, she had endured much more than the average child, yet her classmates remember her as someone who always smiled and always looked on the bright side, even when she was in pain.

Sabrina was a trouper. Though her time with us was all too short, she still managed to give us tremendous joy and to teach us so much. She taught us what love was all about, and she showed us how precious life is.

Born in Montreal, Canada, Sabrina enjoyed watching hockey at a young age with her dad. Every Saturday night, she and her dad had a date to watch the Montreal Canadiens play.

Our family moved to upstate New York when Sabrina was 3. She attended Clover Patch Preschool and moved on to Rosendale Elementary School. In kindergarten, she made a group of special friends who treated her like one of the gang. One of their biggest thrills was to push Sabrina around the school in her stroller/wheelchair.

Sabrina was always a flirt and a busybody, and tended to attract attention. Her glorious curls and big brown eyes didn't hurt, either. She never let her disability get in the way of making friends. She touched many lives.

As we try to cope with losing Sabrina, we want to thank, first and foremost, our parents, brothers and sister-in-law for all their love and support. Thanks also to all of our neighbors, who make us feel like family.

We are also grateful to the staff, parents and students at Rosendale and Birchwood Elementary schools for all they did to honor Sabrina's memory. Thanks to Sabrina's instructional aides. Their dedication was a big reason Sabrina loved school. Thanks also to Niskayuna School District for providing for all her needs.

Special thanks to Brian Alyward for his moral support and for making Sabrina laugh; to the bus drivers and aides who made her rides to school fun; and to our wonderful community who continue to help us through this very difficult time.

We are especially grateful for the kind words that Brian Alyward, Linda Cross and Taylor Tear spoke at the services for Sabrina. Last but not least, thanks to all our extended family and friends for their emotional support.

Sabrina was a lucky girl to have known all of you.

She leaves a large hole in our lives that will never be filled. But she will live forever in our hearts. Her family — Mom, Dad, her sister, Alyssa, and her dog, Emmy—miss her dearly but will cherish her memory always.

NBIA Disorders Association and the Barbiero family thank all those who made donations in Sabrina Barbiero's memory. Such an outpouring of support is truly remarkable and you have our deepest gratitude for your generosity.

In Memory Of Sabrina Barbiero

Brian & Megan Adams
Alda Alves & Lino Pereira
Gaby & Herman Alves
Adilia & Antonio Amaral
Amaral family
Analytical Group/Schenectady Int.
Brian & Sandra Apkarian
Andre & Linda Barbiero
Anthony Barbiero
Ferdinando Barbiero
Giulio Barbiero
Salvatore Barbiero & family

Norman & Marisa Barnes
Linda, Oakley & Ashleigh Bathrick
Dr. & Mrs. Charles Baumgartner
James & Kathleen Baxter
Barbara Belcher
Bruce Belcher
Birchwood Elementary School
Birchwood School PTO
DE Blandy
Jack & Linda Boidman
Matthias Breidsprecher
Vince Bruzzese

Thomas & Bonnie Buckley
Robert & Lauren Burke
Frank & Kim Byrnes
Lillian Cady
Arlindo Calado
Carmille Calado
Capital Care Medical Group, LLC
Capital District Football League
Ronald & Linda Cary
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Ian & Tania Coulthard

Pete Czerpak
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Joan Dinicola
Stephan & Ellen Dirienzo
Anita & Roger Dowse
Richard & Evelyn Dreyer
John & Dawn Dworak
Debbie Ellinger
Epilepsy Foundation of
Northeastern NY

In Memory Of Sabrina Barbiero

Annunziata Esposito	Rosemarie T. Kent	Frank & Felicia Mirolla	Sampson's Prosthetic Laboratory, Inc.
Maria Esposito & family	Rosemary Kolegraff	Pasqualino Mirolla	Daniel & Amy Santandrea
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Timothy & Christine Feeney	Annette Lewis	Nancy & Francis Nappi	Schenectady Int., Inc.
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John & Susan Feyrer	Fatima Magalhaes	Niskayuna Central School District -	Scott & Janice Smith
Claire Gadova	Make-A-Wish Foundation of	Student Support Services	Victoria Stapleton
Cheryl Gerstler	Northeast NY	Donald & Madeline Norman	Daniel & Susan Stec
Eden Gervasio	Gary & Dorilee Male	Nancy O'Donnell	Paul & Roberta Steele
Charles & Judith Griswold	Chantal Malenfant	Mary Olsen	Robert & Patricia Sweeney
Kenneth & Jennifer Grupe	Lisane Manni & Gerry Mirolli	Sharon Olshefsky	Peter & Kim Taormino
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Mr. & Mrs. R. Jaco	Mark & Renee Michalisin	Christine Querbes	Dick & Ann Wyles
Richard Jean	Roberto Michieletto	Christopher Roberts	Robert & Diane Yoder
Kate Johnson	Melanie Miguel	Christine Rockwell	Benjamin Zeller
Philip Johnson	Susan A. Miley	Rosendale School PTO	
Bernice Kane	Mr. & Mrs. Larry Miller	Mr. & Mrs. T. Rossi	

Board

(continued from pg. 1)

away from the disease in March. Gerry and his wife, Anabela, spearheaded a fundraiser last August that netted \$23,000 for our Research Fund, plus a \$50,000 grant from the Wright Family Foundation for research equipment. The association used this grant to become founding members of the Genetic Alliance Bio Bank.

Gerry has worked since 1998 as a research chemist for Schenectady International, a family-owned chemical manufacturing company in upstate New York. He leads a group focused on developing new types of polymeric walls to meet commercial industrial needs.

Gerry received a bachelor's degree in chemistry, a master's in electrochemistry and a doctorate in organic and polymer chemistry.

Born in Montreal, Canada, to Italian immigrants, Gerry's major interests are skating, woodworking, gardening, biking, reading technical journals and books and spending time with his family.

Our third new board member is Phil West from Vancouver, Wash., which is just five minutes outside of Portland and near the Oregon Health & Science University, where research into NBIA is being conducted. Phil is the brother of board member Kris McGourthy and the uncle of two boys with NBIA.

Phil grew up in Mountain View, Calif., and received a bachelor's



Kris McGourthy and her brother, Phil West, a new board member for NBIA Disorders Association.

degree in business from Oregon State.

He has recently accepted a job working for Microsoft in sales. Before that, he worked for Tripwire Software, selling security software to the government and he also worked for Hewlett Packard in Washington, DC.

Phil has experience as a secretary for an education non-profit. He is eager to lend his expertise to our group.

He loves to play golf and snowboard in the winter. He also enjoys traveling.

The other board members, in addition to Tapke and McGourthy of Middleboro, Mass., are President Patty Wood of El Cajon, Calif.; Susan Laupola of Cincinnati; and Mary Ann Roser of Austin, Texas.

Board attends Genetic Alliance Conference for the first time

By Patricia Wood

For the first time, the entire NBIA Disorders Association Board met face-to-face and attended the annual conference of the Genetic Alliance, an organization that is becoming increasingly important to our group. The alliance provides advocacy, information and technical guidance to lay advocacy organizations, and we are now counted among its 600 members.

The conference was an opportunity to learn about cutting-edge technologies, to network with fellow advocates and to be inspired by the stories of others who have accomplished the seemingly "impossible." The groups that make up the Genetic Alliance are united in their belief that there are many common threads tying them together, giving them power as one voice.

Our association is also a founding member of the alliance's Bio Bank, which is a repository for blood and tissue samples owned by each group. We are still in the set-up phase of the Bio Bank and have not yet started collecting samples.

At the conference, all NBIA board members attended workshops on how boards can work together more effectively. Other seminars attended focused on fundraising, partnering with industry, foundations, and government organizations that work with rare disease groups.

As one board member said: "I was amazed at the information presented and the open discussion between the advocacy groups. We need these conferences to teach us new changes in government policy, how to work with industry to advance research, and to generate funding for our disorders. Most importantly, they help provide the emotional strength needed to achieve our goals."



NBIA Disorders Association Board - First Row: Mary Ann Roser, Mary Tapke, Phil West, Patty Wood, Susan Laupola, Kris McGourthy
Second Row: Gerry Barbiero, Jeff Doerner

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

Alyssa Barbiero
Norman & Marisa Barnes

Becky Belcher
Lois Clifford
Mrs. Marion Hermann
G. Yvonne Sweet

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Jillayne L. Kuz
Tara Quenneville

Brent Bonfiglio
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Michael Knowles
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Pete Cassidy Family
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McGourthy Family
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Nancy Overman

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Christopher Pichardo
Betty Castillo

Julie Smyth
Maxine Smyth

Ken Stromsta
Arleen Meyer

Kimberly Wood
Don & Marjorie Cullen
Ron & Jill Kosen
Charles & Diane Murdock
Regina Neal

In Memory Of

Alexander Kissling Diaz
Joan Kissling

Alfred Bernard Donahue
Joan Saunders

Amy Elizabeth Hall
Yvonne Hall

Clarence Roser
Anella Roser

Jean Venuto
Gary & Carol Steinker

MESSAGE FROM THE PRESIDENT



Patty Wood

Our family marked a milestone this summer: My daughter, Kimberly, turned 18 on June 25. We had birthday parties starting the week before, beginning with a Hawaiian party at her Dad's and stepmom's house. On her special day we had a cook-out at the beach with a bonfire, and then watched fireworks from nearby Sea World. Throughout, we made sure to gather those close to us to celebrate her life these past 18 years.

On reflection, we stopped to realize how many have played a big part in our lives and who have helped us along the way. We've had caring teachers at every step of our journey. An aide is with her every day at school, taking care of all of her needs so she can mainstream with the other students as much as possible. We've had many wonderful nurses come into our lives—and into our home—to help us take care of Kimberly.

Then, there's the physical therapist who works with us to keep Kimberly as comfortable as possible and to move her body to maintain as much mobility as possible. We can't forget the speech therapist who has worked diligently to find a way for Kimberly to communicate using switches and buttons and anything else that might work! We fondly thank our occupational therapist who recently moved on and will be sorely missed. She was so creative in thinking of ways to solve problems and so mindful of Kimberly's unique needs. The hand splints, headrests and many other innovations we tried over the years are a testament to her dedication.

There have been many doctors over the years, including several who have hung in with us from the very beginning. The pediatrician who greeted her in the hospital when she was born is still seeing her today. Her neurologist and orthopedic doctors have been with us since the disease began and have been a constant source of information and caring help.

The doctors and staff at the Osteopathic Center for Children in San Diego have been helping Kimberly since she was 2. She made weekly visits for many years and now we go bi-weekly to help maintain her body as best as possible. And, of course, there are many other doctors we call upon from time to time, including the gastroenterologist, the ears, nose and throat doctor, the pulmonary specialist, the metabolic specialist and many other specialists who have answered our many questions and whom we have learned from over the years. Thanks to the diligent, inquisitive researchers who are working on this disease. We hope you never give up!

We have received so much help and for that we are very grateful. Thanks to everyone who has been a part in Kimberly's life – who has made her 18 years special. Never has the adage, "it takes a village to raise a child" been more true. We couldn't have done it without each of you.

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.

Ben & Jerry Foundation
 Slava & Maria Buchkovich
 Paul & Janet Buhay
 Karen Campbell
 Wayne Gateman
 James Hayes
 Jennifer Hide
 Mr. & Mrs. George Jones
 Mr. & Mrs. Lowell Judd
 Genghis & Nahla Khan
 Kathleen Kindregan

Kristine Klein
 Ella Laird
 Richard & Jeanne MacDonald
 Michael Maitin
 June McClure
 Mr. & Mrs. David Miller
 Jerry Oshiyama
 April Pendera
 Mr. & Mrs. Oscar Reeder
 Joan Taylor
 Michael J. Vince, III

NBIA Disorders Association would like to thank the **Glen Wright Donor Advised Fund of the Greater Cincinnati Foundation** which awarded a \$2,000 grant to our organization in May.

Your support is greatly appreciated.

Wedding Day Gift

The NBIA Disorders Association thanks Brian and Megan Adams who made our organization a part of their wedding day. The couple chose to make a donation to the association in lieu of giving traditional favors to the guests at their wedding.

This gift was made in memory of Sabrina Barbiero. We deeply appreciate the couple's generosity.



Patty & Kimberly at Ski Beach in San Diego on her 18th birthday.



2082 Monaco Ct.
El Cajon, CA 92019-4235

Formerly
Hallervorden-Spatz
Syndrome
Association



Our Mission:

NBIA Disorders Association is a non-profit organization dedicated to providing emotional support to families affected by NBIA, educating the public about this disease, and monitoring and supporting research and informing others of its progress.

NBIA Disorders Association

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Visit our Web site at

www.NBIAdisorders.org

from discovery to cure

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