

Board awards three \$30,000 grants to study NBIA in 2007 grant cycle

By Patricia Wood

The NBIA board recently chose three exciting proposals to receive \$30,000 each for research into NBIA disorders. The recipients are Dr. Susan Hayflick of Oregon Health & Science University, a familiar name among those receiving NBIA grants, and two first-time grant honorees, Dr. Paul Kotzbauer of Washington University and Dr. Soma Das of the University of Chicago.

These awards bring to 11 the grants the board has made in the past six years. They once again demonstrate the power of family fundraisers and private donations that make these grants possible.

These grants often help scientists compile preliminary research information so that they can obtain further funding from sources such as the National Institutes of Health. With NIH funding becoming more scarce, these grants are increasingly important to ensure that research into NBIA disorders continues.

Dr. Susan Hayflick has received six grants in six years from the NBIA board. Her project, "Towards Clinical Therapeutics in PKAN" builds on 15 years of research advances.

"Our family studies led to the gene's discovery, which in turn led to understanding the biochemical and

(see grants on pg. 5)



OHSU lab participates in interdepartmental pumpkin carving contest.

Pictured: Wei-Hong Xiong, Moon Yoon, Thuy Nguyen, Allison Gregory, Susan Hayflick, and Brenda Polster.

Not pictured: Penny Hogarth

Do you have a special skill or know someone who does?

At 11 years old, the NBIA Disorders Association is ready to take a big step forward.

But it can't do it alone. The board has seven committed board members with a wide variety of skills, but it needs more hands to accomplish what it wants to do in the near- and long-term. At a recent meeting, the board decided that it needed, first and foremost, to add board members with specific skills and to add volunteers and new programs so it could better meet the needs of the individuals and families it serves.

The board is asking that anyone who has the talent, skills or

motivation to assist in this effort, to step up to the plate. Some of you may have friends or relatives who have asked how they could help and have the time and energy to devote to this cause. Here's a chance.

(see board 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-profit in Germany who works with us in the promotion of research and treatment of NBIA, can be contacted at the following address:

Hoffnungsbaum e.V.

Hardenberger Str. 73

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Web.: www.hoffnungsbaum.de

E-Mail: hoffnungsbaum@aol.com



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

Board

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Specifically, the board is looking for people with talents in fundraising, grant writing, organizational leadership, non-profit expertise, strategic planning and other skills that would help us move forward.

With the right people in place, we hope to achieve the following:

- *Create and finance a fellowship program that would encourage more researchers to take on our disease.
- *Develop an outreach program that would ultimately have every person affected by NBIA on our registry.
- *Raise enough money to hire a small paid staff. We need a full-time executive director and at least a part-time clerical assistant as soon as possible.

So many of you have been creative at putting on fundraisers, and we urge you to keep it up and hope others will join in. We also are eager to cultivate a cadre of volunteers we can call on to help us serve families more effectively and make our organization better. Do you have some skills you'd like to share with other NBIA individuals or families? Do you know of some service we should provide that we are not providing? If so, please let us know what you're willing to do and what we should be doing that we are not doing.

We would like to create programs that a volunteer director would be responsible for handling and who would report to the NBIA board. Some ideas we are considering are:

- * A bereavement program for families that have lost loved ones.
- * A program to welcome newcomers with regional coordinators.
- * A conference planning program.
- * An education and awareness program to coordinate accurate, up-to-date information on NBIA for media, physicians and other interested parties.
- * A community fundraising support program that would work with families wishing to hold fundraisers in their communities.
- * A physician referral program that would generate a list of neurologists and neurosurgeons that have familiarity with NBIA to help families find a physician.

Anyone interested in helping as a board member or the volunteer director of a program is asked to forward their [resume and letter](#) for consideration to Patty Wood at pwood@nbiadisorders.org. NBIA board members also are volunteers and are not compensated for their time.

Thanks for your help!

NBIA Disorders Association Board of Trustees

NBIA Disorders Association says adieu to two board members; elects new vp

The NBIA Disorders Association bid farewell to two of its nine board members this year and elected charter board member Susan Laupola of Cincinnati vice president.



*Gerry Barbiero
of Niskayuna, New York*

The board thanks Gerry Barbiero of Niskayuna, New York, and Lisa Shook of Cincinnati, Ohio for their service. Gerry joined the board in 2004 and became vice president in 2005. He and his wife Bela were vital members of our organization and worked tirelessly to promote awareness of NBIA, raise money for research and generate support for the organization.

The couple had two girls with NBIA, Sabrina and Alyssa, who passed away in 2004 from the disease. Gerry and Bela will be missed.

Lisa joined the board in early 2006 and came up with the idea of creating a cookbook to celebrate the organization's 10th anniversary. The cookbook proved to be a major fundraising success.

Susan has handled numerous duties for the board since its inception, including its incorporation papers and tax filings in the various states. She was the board's secretary for several years and has shown strong leadership throughout her service.



*Lisa Shook and Susan Laupola of Cincinnati, Ohio,
and Bela Barbiero of Niskayuna, New York*

NBIA: who's under our umbrella?

By Allison Gregory

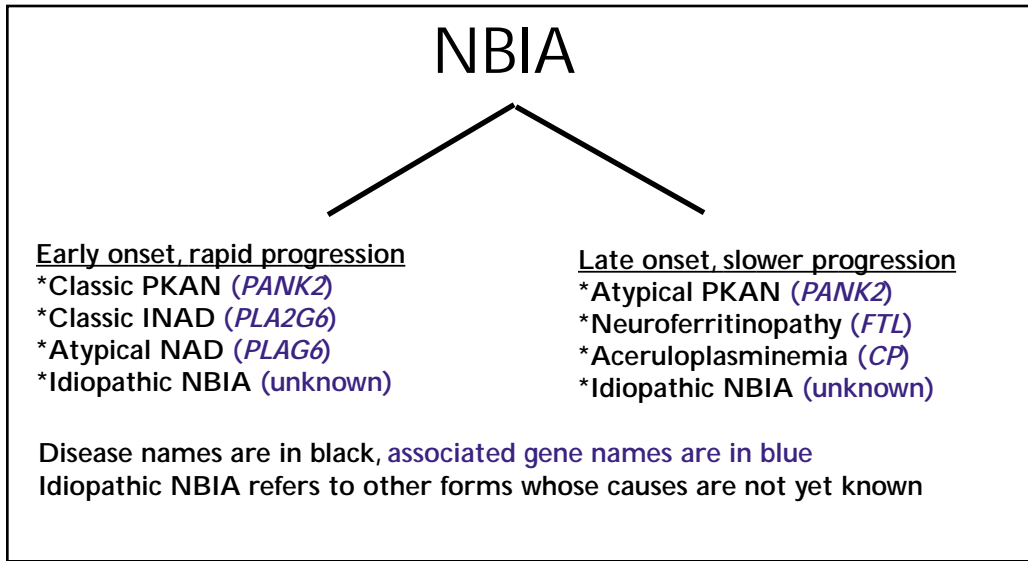


Allison Gregory, Genetic Counselor at Oregon Health & Science University

When a family first receives a diagnosis of neurodegeneration with brain iron accumulation, or NBIA, they soon realize that part of the challenge of learning more about this group of disorders is simply understanding all the names and related acronyms. This may be a challenge that their physicians take on as well, since historically NBIA was called Hallervorden-Spatz syndrome and this is still a name that appears in many medical texts and journal articles.

As we learn more about the group of disorders that involve neurodegeneration and accumulation of iron in the brain region called the basal ganglia, the related language has also evolved to reflect our new understanding. Identification of genes causing different types of NBIA has also helped us to better categorize the different disorders falling under this umbrella. The term NBIA is general enough to cover all conditions previously categorized as Hallervorden-Spatz syndrome plus other conditions found to fit in this group.

The diagram below shows the different forms of NBIA and, when known, the genes that cause them. Although NBIA is generally divided into early onset and late onset forms, there are always exceptions to this rule and some cases will fall between these two categories.



Most recently, we have proposed to add classic infantile neuroaxonal dystrophy (INAD) and atypical neuroaxonal dystrophy (NAD) to the NBIA umbrella. These are both caused by changes in the *PLA2G6* gene. The Hayflick research group initially started studying INAD because several physicians worldwide sent us examples of patients with this diagnosis who had brain iron accumulation identical to that seen in NBIA. At first, we thought these cases were rare, but we have since found that several individuals with INAD also have high brain iron in a similar pattern to other NBIA.

In addition, individuals with this diagnosis have axonal spheroids like

those seen in NBIA, although in INAD they are not limited to just the brain but can also be found in other tissues like skin or muscle. We also found that some individuals who we never suspected to have INAD have mutations in the *PLA2G6* gene. Unlike classic INAD, which usually involves floppiness (hypotonia) in infancy, followed by spasticity (stiffness), these atypical individuals have later onset with dystonia as a main symptom. For this reason, we created a separate category called atypical NAD.

Two other rare disorders are also considered to be part of NBIA, although to date they haven't received much attention within our NBIA community. This is primarily because they affect small, discreet groups of people and appear to be even more rare than PKAN or other types of NBIA that many of us know well.

Aceruloplasminemia has mainly been studied in Japan, where it affects about 1 in 2 million adults. It is unclear how often it occurs outside the Japanese population, but is even more rare. Aceruloplasminemia has three main symptoms: retinal degeneration, diabetes, and neurologic disease. These are caused by iron accumulation in the brain and visceral organs. This is a disorder of adulthood that usually

(see *NBIA* on pg. 5)

NBIA

(continued from page 4)

begins during the third to fifth decade of life. Like most NBIA, it is inherited in an autosomal recessive fashion.

Neuroferritinopathy typically starts in adulthood with dystonia, jerky movements (chorea), and mild changes in thinking (cognitive effects). The prevalence is unknown, but fewer than 50 cases have been described in the literature and most of these individuals have the same gene change, suggesting a common ancestor. Brain MRI shows abnormal iron accumulation in the basal ganglia during early disease; later, cysts develop in these regions. So far, neuroferritinopathy is the only type of NBIA known to be passed through the family in a dominant pattern, meaning that an affected individual has a 50% chance of passing it to each of his/her children.

Of all cases of N B I A , about one-half to two-thirds are accounted for by one of the conditions described above... the remaining individuals are said to have "idiopathic N B I A , " meaning that the underlying cause is not yet known.

Of all cases of NBIA, about one-half to two-thirds are accounted for by one of the conditions described above, with PKAN being the most common. The remaining individuals are said to have "idiopathic NBIA," meaning that the underlying cause is not yet known. For many of these families, the person diagnosed with NBIA is the first and only affected individual, so it is difficult to know whether there is a specific pattern of inheritance.

It is thought that most of these cases are probably recessive because (a) there are some families with more than one affected child and (b) idiopathic NBIA is more common in families where the parents are related, such as distant cousins (this makes it more likely that they share a common recessive gene). The symptoms in this group are more varied because there are probably several different causes of neurodegeneration in this group. As with other forms of NBIA, there are both early-onset and late-onset types.

As we and other researchers continue to identify genes that cause NBIA, it is likely that definitions will continue to change. We hope that expanding this NBIA umbrella and trying to understand the underlying processes that are common among people with different forms of NBIA will lead us to new treatments and a better understanding of this disease.

For more information on this topic, please visit our Web site at www.NBIAdisorders.org, NBIA FAQ link, "What is NBIA?" which has been updated to reflect new information.

Grants

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cellular changes caused by mutations in *PANK2*," Hayflick said. Those new understandings of the changes allows Hayflick to test therapies in a mouse model of PKAN developed by collaborator Jane Gitschier's team at the University of California, San Francisco.

"With Penny Hogarth's expertise in clinical therapeutics, our OHSU/UCSF research team is poised to perform the first drug trial in these mice," Hayflick said. "Additional animal drug trials will follow, with the goal of moving these to human studies as soon as we find a promising agent. Although the mice do not show dystonia or brain iron accumulation, they have key disease characteristics similar to those in PKAN patients that enable these studies. This project marks the beginning of an important phase in NBIA research as we move 'from discovery to cure.'"

Dr. Paul Kotzbauer's project, "Combined disruption of mouse *PANK2* and *PLA2G6* genes to generate an improved animal model of NBIA," said his goal is to develop a mouse strain that has the central features of NBIA.



*Washington University Lab
Mengyang Sun, Ibrahim Malik, and Paul Kotzbauer*

"An improved mouse model of NBIA would enhance our ability to determine whether a particular therapeutic approach is likely to be helpful and guide decisions to pursue further studies in clinical trials," he said.

Lab, or "knockout" mice that have mutations in one of the two NBIA genes discovered so far, *PANK2* or *PLA2G6* genes, each reproduce some aspects of the disorders but lack the changes common to all forms of NBIA, which include iron accumulation and neurodegeneration in a brain region called the globus pallidus, Kotzbauer explained.

(see grants on pg. 11)

2nd Annual Queen Bee Golf Marathon raises \$41,500 for NBIA research fund

By Rich Leap

We learned so much from our first golf marathon that we thought the second one would be much easier and contain fewer surprises. While that certainly was true for the planning, we were pleasantly surprised by the overwhelming support from golfers and supporters who came out in October to compete at the Piedmont Country Club in Northern Virginia.

Quest Diagnostics came on board again as the marathon's sponsor. That ensured that 100 percent of the pledges went to the NBIA Disorder Association's research fund. We also obtained enough prizes through local sponsors to keep the play interesting and ensure that none of the players would leave empty-handed.

Our goal was to beat last year's event and raise \$30,000.

Twenty-five players (23 men and two women) arrived with pledge sheets in hand. (Two other golfers, Mike Leap and John Moore, raised funds, but were unable to play this year). We had a picture-perfect day, cool to start and warming up to the mid-70s.

My daughter, Brittany Leap, who has NBIA and in whose honor the fundraiser was held, arrived on the course to visit with all the players and encourage them. She loved riding in the golf carts while being greeted by each group.

By the end of the day, 2,262 holes of golf had been played. Eleven golfers reached the 100-hole goal, with the average being about 90 for the day. The day was capped by dinner and awards as a tired but supportive group vowed to compete again next year.

Because of the tremendous work by golfers in gaining sponsors, we easily surpassed our goal, raising more than \$41,500 for NBIA research. The support for Brittany, the event's Queen Bee, and NBIA continues to amaze us.

We have so many people to thank: Peg Sherwood, for creating and donating the Hole Sponsor signs; Thom Wood for taking photos all day; the golfers, sponsors, volunteers and prize donors. All of their efforts help lead us closer to a cure.



Golfers Pictured: Chris Prestera, Kevin Humphrey, Rich Erikson, Rich Leib, Mike Knapp, James Penn, David Waits, Florinda Russell, Rich Leap, Frank Wood, Brenda Godwin, Kevin Rollison, Neil Russell, Jay Portnoy, Tom Huard, Chris Kase, Mitch Russell, Chad Cutlip, Sid Vikram, Jim Nix, Michael Menn, Dean Updegrove
Not Pictured: Bryan Firestone, Rob Seamen, Rick Phillips

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*Volunteers Pictured: Samantha McDade, Teresa Markham,
Heather Porter, Peggy Gough, Robin Firestone
Not Pictured: Nadine McDade, Judy Leap, Michelle Ayala*

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Classmates of Ben Patterson remember him with bracelets



Hailey Lachowsky, Michala Roberts, Anna Standridge, Austin Tate, Robbie Powell and JP Bradley show off their "Live 4 Ben" bracelets being sold to raise money for NBIA research in memory of their friend, Ben Patterson.

Students at Bob Courtway Middle School in Conway, Ark., have found a way to memorialize their schoolmate, Ben Patterson, who died Aug. 14 after a 12-year battle with NBIA. They have been selling "Live 4 Ben" bracelets to raise money for NBIA.

The effort has been spearheaded by Patterson's friends, Robbie Powell and Michala Robertson. Robbie said the bracelets are green, in honor of Ben's favorite color.

The students are selling the bracelets to their friends at school and to fans who attend home football games. So far, this special group of students has raised \$1,500 in their friend's honor.

Jewelry party fundraiser nets \$440 for NBIA fund

By Kristi Ose

Here's an easy way to raise money to benefit your favorite charity: the NBIA Disorders Association.

My friend Julie McGovern, of Goshen, Kentucky, hosted a Silpada sterling silver jewelry party in honor of my son Jared who is six years old and has INAD, a form of NBIA.

Johnna Borgmeier, the jewelry representative, agreed to donate 15 percent of the total retail sales to the NBIA Disorders Association.

Dear Ms. Wood,

Last year, my brother Biaggio (7), my sisters Elly (9) and Lia (3), and I, Josie (11), decided that instead of gifts for our birthdays, we would ask people to donate to NBIA instead. It went so well, that we did the same thing this year.

We are thrilled to send \$700 for your continuing research into finding a cure for this disease. We know that \$700 could buy a lot of toys and presents, but you see, we also know Brittany Leap (who has NBIA). Because she is our very good friend, we know that no present is more valuable than her presence. So, we are happy to be able to do something to help someone who means so much to us.

Thank you for everything you do in your work to find a cure for NBIA. May God continue to bless you.

Sincerely,

Josie, Elly, Biaggio, and Lia Corrado
Gainesville, Virginia



Biaggio Corrado, Brittany Leap, Elly Corrado and Josie Corrado. The Corrados live in Gainesville and Brittany lives in Haymarket, Va.

That donation amounted to \$225, and others gave direct contributions to the organization, making the grand total \$440 for the event.

The amount raised exceeded our expectations since only 12 people attended the party. We are grateful for the generosity of everyone who helped make this event so successful.

Anyone interested in doing a party like this for a fundraiser should consider talking with a home sales representative to see if they would be willing to donate part of their sales proceeds to the NBIA Disorders Association. For a smaller scale fundraiser, it required little work and planning and was a fun way to raise awareness and money for the organization.

Family Masquerade Ball nets \$2,500 for NBIA

By Josie & Elly Corrado

We got into the Halloween spirit and ended up raising \$2,500 for NBIA by finding a fun way to support our friend, Brittany Leap of Haymarket, Virginia who was diagnosed with NBIA in 2006.

The money will go to NBIA's research fund to help find a cure for the disease.

The event was a Family Masquerade Ball held Oct. 20. Not only did it feature a costume party for the entire family, it included a DJ, dancing, a craft table, games and a raffle. Raffle prizes included something for everybody: Dolls, books, DVD's, a DVD player, restaurant gift certificates, games, dental services, goodies for golfers, and our "big" item was a signed script from the cast of "Hanna Montana!"

We also designed T-shirts and bracelets to sell, and Father Peffley of Holy Trinity Catholic Church demonstrated his talent for juggling bowling balls and even machetes!

The Methodist Church in Gainesville donated its hall for the event, and many local businesses donated prizes. Food came from Papa John's Pizza, Harris Teeter and Bloom Grocery stores.

The entire Corrado family was involved in the project: parents, Colleen and Rick, along with Josie, 11; Elly, 9; Biaggio, 7; and Lia, 3.

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Masquerade Ball in Gainesville, Virginia organized by Josie and Elly Corrado.
Front row: Brittany Leap, Lia Corrado
Back row: Elly Corrado, Josie Corrado, Biaggio Corrado

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Kris West, a gym instructor at Birchwood School in Niskayuna, New York, surrounded by students at a "Family Fun Walk" fundraiser. \$1900 was recently donated from a portion of the proceeds to NBIA Disorders Association in recognition of the positive impact the organization had on the Barbiero family, and in memory of Sabrina and Alyssa Barbiero.



Students at the "Family Fun Walk" in Niskayuna, New York .

Grants

(continued from page 5)

"We speculate that the *PANK2* and *PLA2G6* genes have overlapping functions in the brain and that although brain cells in mice may be able to compensate for the loss of one gene, disrupting both genes together may be sufficient to cause a disorder in mice that is more similar to what occurs in humans," he said. "We will breed mice from the two existing knockout strains over several generations in order to generate mice that have mutations in both the *PANK2* and *PLA2G6* genes. We will then look for features of NBIA using tests for neurological function and microscopic examination of brain tissue."

Dr. Soma Das's project is "Deletion and duplication analysis of the *PANK2* and *PLA2G6* genes in patients with neurodegeneration with brain iron accumulation," which could improve the diagnosing of NBIA patients.

Das explained that some patients with NBIA clinical symptoms may not have had any mutations in the *PANK2* or *PLA2G6* genes identified and their diagnosis remains uncertain.

While "DNA sequencing has been the traditional method of mutation detection/testing in patients with NBIA disorders, certain types of mutations, such as deletions and duplications within the *PANK2* and *PLA2G6* genes will be missed by DNA sequencing," Das said. "We will use some newer methods of DNA analysis that will pick up these types of mutations. We may therefore be able to identify mutations in patients in whom no mutations have been identified to date. Our studies should result in the development of a more comprehensive test for NBIA disorders."

Our organization is excited about these new projects and the potential they have to enhance our knowledge of NBIA disorders. The research program, and the support families provide it, is the vital link to the organization's quest for a cure.



*University of Chicago Lab
From left to right: Soma Das, Melissa Dempsey, Lorena Diaz deLeon, Anthony Lozada, Roshni Alva, Eden Haverfield, Amanda Reeder, Peixian Chen, Shalini Reshmi and Lucy Liu.*



Benjamin Scott Patterson

November 21, 1994 - August 14, 2007

Brother 'Bubby' and a friend recall Ben Patterson's remarkable life

By Will 'Bubby' Patterson (in italics) and Scott Maple (in regular type)

Take it from his "Bubby," Ben Patterson had a way of putting things into proper perspective.

You were a lover, not a fighter. You smiled through all of the things that happened to you, which is more pain than I will probably ever go through.

With all his body had to endure, it would have been understandable if Ben had withdrawn. But he had too much to offer. To Will, his older "Bubby." To his parents, Bill and Kim. To his grandfather, Ray. To his special-needs teammates on the Conway Braves baseball team. To those he warmed with a smile at church or school.

Without you my life would be so much different. You taught me not to give up, to keep fighting and to keep smiling.

It was tough to watch as NBIA erased the mobility Ben had so enjoyed. No more running full-tilt, bouncing off walls and bearing the scars that marked him as all-boy. Yet being at the mercy of others to serve as his hands and feet didn't keep him from romping with Will.

You had my back no matter what, and I always had yours. You did much more for me in 12 years than most people will do for me in a lifetime. I am blessed to have had you for so long.

Blessings flowed in all corners of Ben's world, including for friends made at school in Conway, Ark. Especially Robbie Powell, his best pal from first grade on.



Kathleen Galt

June 8, 1992 - September 17, 2007

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

You were always there for my brother and deeply loved him. You would come over to the house and sit all day even if he was asleep, and those were the times I could tell you truly cared.

Friends also saw Ben's precocious side — including his flirtations with girls who would skirmish over who got to wheel him to recess. (The kisses that ensued would occasionally get him in trouble with his teachers.) His charm carried over into his home life, as well, where his granddad Ray — Ben called him Papa — was putty in his hands.

You impacted his life more than anyone, and he impacted yours, I know. When we were in a room and you walked in he would completely ignore whatever was happening and concentrate on you, his favorite person.

Now he will focus his charms on dear "Gram," who preceded him and Papa in death. And on his Heavenly Father, who made a special place for him free of pain.

It makes me so happy to know that finally you have gotten what you deserve. You have gotten peace. Peace in mind and body.

And those left behind have gotten so much as well — and not just fond memories: There's also hope. There's persistence. There's faith and love. And an example of how to live — each day.

I truly owe my life to you and will try and live it the way you would. You will always stay with me, leading me, and I thank you so much for that. I know in my heart that you're just as much here now as you ever were.



Kenneth Peter Stromsta
October 31, 1986 - September 14, 2007

Kenny was truly a gift from God. We adopted Ken at 10 days old and he immediately captured the hearts of all who met him from that day on. His dad says he had a winsome smile that would melt your heart. When he was just 4, Ken won the Mr. Personable award at school.

Even in the mist of daily pain, Ken lived a life of purpose. This was something we wanted people to know about those with special needs. His neurologist said that was one thing Ken had taught him and "much" more.

Most people remember Ken for his sense of humor and how he liked to tease. But we remember him for his unconditional love, his super hugs and unbelievable strength. We remember how he laughed so hard at his dad, he could hardly breathe; how he would come home from school with kisses all over his face; how he loved to bake cookies with his sister and listen to music in his brother's room; how he would play Pretty Pretty Princes with dad; and how he would enjoy those long walks with mom.

But we also remember the times of great pain. His biggest comfort was when we talked about heaven and promised some day he would walk and talk and never have pain again. He would smile so big and we could see in his eyes he believed without doubt. And now we feel we have kept our promise.

We lost more than a son and brother, we lost our best friend, our buddy. We believe if Ken could have talked, his last words would have been, "Mom, tell everyone, thank you."

We love and miss you more each day.
 Always,

Mom, Dad, Laura & Jeff



Brent Michael Fry
June 30, 1989 - September 21, 2007

Brent was a very loving and happy little guy. We are heartbroken that he has left us. We know his pain and suffering are over and that he is with God, but our hearts will ache forever. His Papa Jordan said it best: "Brent never said a bad word about anyone, Brent never did anything mean or hurtful to anyone. He always had a twinkle in his eye and a smile on his face." Brent was indeed an angel, and he touched everyone's heart. He will be forever missed by those who loved him.

A Parent's Love

We didn't have to look into your eyes to fall in love with you. We didn't have to hear your cry to know you loved us too. We didn't need to hold your hand to cherish you always. You touched our souls. You sweetened our spirits. You gave us memories we'll always hold dear. Yes, our hearts ache because you departed us too soon, but a parent's love does not end with death, for you are our child, and our love forever is yours.

Melinda & Mike Fry
 Papa & Mema Jordan

*When you are sorrowful look again
 in your heart, and you shall see that in
 truth you are weeping for that which
 has been your delight.*

Kahlil Gibran

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.

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Becky & Barbara Belcher
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Kathleen Galt

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In Memory Of

Alyssa & Sabrina Barbiero

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Andrew & Holly Stadel
Bob & Pat Sweeney

MESSAGE FROM THE PRESIDENT



Patty Wood

This summer and fall were hard for NBIA families. We lost four children in just over a month, three of them in one week. They all belonged to families active in the organization, families I know well. I have e-mailed with all of them over the years; two were at our family conference in May.

Pam Stromsta was the first mother I met who had an NBIA child when I was searching for other NBIA families for two years after Kimbi was diagnosed. Kenny was the same age, and he shared a common history of the disease with Kimbi. We stayed close over the years and supported each other in good times and bad.

Even as our circle grew over the years, somehow I felt as long as Kenny was here, Kimbi was safe, too. They had both made it through many close calls and I thought that would continue for many more years. Then, suddenly, Kenny was gone and the reality of what this disease does hit close to home. I felt very vulnerable, like every moment is precious and we don't know when the day will come and our kids will just not pull through.

I now know of only one other child who has lived with the severe form of the disease from a young age who is older than Kimbi — by one year. This is not a contest I ever wanted to join or to win. I want all of our NBIA children to make it into adulthood and not have to suffer the pain and discomfort so many have.

When Kimbi was young I used to think I could not cry because I might not ever stop. But I learned we have to grieve, and then go on. Now every time I hear the sad news of someone's passing, I cry and mourn for the family and say my prayers they will make it through their difficult time. And then I get back to the organization's work, realizing that maybe what we are doing will help families in the future not have to endure this wrenching pain.

Perhaps that future is not so far away. Some families are starting to turn to Deep Brain Stimulation surgery as a way to slow the progression of the disease and improve the quality of life of NBIA individuals. Many have seen promising results. Just yesterday I heard from a family whose NBIA-affected daughter had the procedure. The day after the settings were adjusted, she started speaking again, for the first time in a year. Improved speech is not even a normal outcome for DBS surgery, so what a blessing that was for this family. I would give everything I own to hear my daughter say "mama" once more.

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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Visit our Web site at www.NBIAdisorders.org to order.

It is vital that we continue to support NBIA research if we want more extraordinary results like this. We need to welcome new researchers into our NBIA family and help them in any way possible to continue to improve the lives of NBIA individuals. We must never lose four of our children in one month again.

As we celebrate this holiday season, let's hope for continued progress and more wonderful success stories in our future.



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Our Mission:

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

NBIA Disorders Association

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