



Bruce Belcher, Barbara Belcher, Ben Belcher, Cheryl Lamos and Becky Belcher (center) from Rensselaer, New York.

Somewhere between disbelief and exhaustion,

New York mother learned to accept and adapt

By Cheryl Lamos

At first, I was in disbelief. How could not one, not two, but three of my children be going through this horrible ordeal? Yet, the signs, slow and subtle at first, crept into the lives of my children, one by one.

Looking back now, I can honestly say that NBIA struck without warning. My children rode bicycles, rollerskated, played sports and did all the things other children did. It wasn't until my oldest, Rebekah (we call her Becky), who will be 27 in May, started showing signs of clumsiness at about age 16 that we thought something might be wrong. The best way I can describe the sign was uncoordinated, awkward walking.

We also noticed that Bruce, who is now 25, had started walking on his toes. Bruce had been diagnosed with attention deficit and hyperactivity when he was in the fifth grade and responded well to Ritalin for many years. Was the hyperactivity a part of NBIA? To this day, we don't know.

Barb, who will be 24 in May, also had a milder form of attention

(see Lamos on pg.4)

NBIA Disorders Association joins Genetic Alliance Bio Bank

By Patricia Wood

Thanks to a grant from the Wright Family Foundation in September, our organization will be on the front lines of an effort that we expect to reap huge benefits for NBIA research. The \$50,000 foundation grant will be used to cover the estimated five-year costs of joining the Genetic Alliance Bio Bank.

This bank will store blood and tissue samples from patients with various disorders. These samples will be used for research purposes, so scientists can learn more about NBIA and other disorders. The ultimate hope, of course, is that researchers will discover cures for the diseases being studied.

If the bio bank lives up to its potential, it can change the way research is being done today.

Patient advocacy groups representing these individuals, such as the NBIA Disorders Association, will own the samples. It is less expensive to have a group form a bio bank, rather than each organization trying to go it alone.

But there also is power in numbers. Together, our organizations can mentor and support one another as we learn more about the illnesses affecting our members. The bio bank also can empower advocacy groups to work more closely as partners with the researchers. The hope is that the bio bank will make the research more streamlined and coordinated so that beneficial treatments can be delivered to patients more quickly.

Often, research into rare disease is hampered because the number of samples being used in studies is very small, too small to give a clear picture of the disease. This can lead to inaccuracies

(see Bank on pg. 6)

Inside This Issue...

California Fundraiser	2
Senator Kennedy visit	3
Manage your medical journey	7
NBIA and morphine	7
Our Generous Supporters	11
Message from the President	11

MEDICAL ADVISORY BOARD

Susan Hayflick, M.D.
Geneticist & Pediatrician
Oregon Health & Science University
Portland, Oregon

Rayburn Skoglund, M.D.
Pediatric Neurologist
Childrens Associated Medical Group
San Diego, California

Kenneth Swaiman, M.D.
Pediatric Neurologist
University of Minnesota
Minneapolis, Minnesota

Ramesh Tripathi, M.D., Ph.D.
Dept. of Ophthalmology
University of South Carolina
Vision Research Laboratories
Columbia, South Carolina

Officers/Board of Trustees

President - Patricia V. Wood
El Cajon, California

Secretary - Debbie Forstall
Northridge, California

Treasurer - Gayle McMahon
El Cajon, California

Board

Dianne Gray
Naples, Florida

Susan Laupola
Cincinnati, Ohio

Mary Ann Roser
Austin, Texas

Kris McGourthy
Middleboro, Massachusetts

Mary Tapke
Cincinnati, Ohio

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.

Family grapples with son's NBIA diagnosis by helping to raise money to battle disorder

By Gaetano and Veronica Bonfiglio

When our son, Brent, was recently diagnosed with NBIA, we wanted to do something to raise funds for research and spread awareness of this disorder. At the same time, we wanted to be low-key about it. We felt it was important for Brent to keep a positive outlook and not be aware of his prognosis, unless it was absolutely necessary.

The end of the year is typically a time for making charitable contributions, and companies, like the one Gaetano works for, Hewlett-Packard, have a matching gift program to encourage employee donations.

We registered the NBIA Disorders Association with the company's charitable giving arm, and we donated to the NBIA association and to Dr. Susan Hayflick's NBIA research group at the Oregon Health & Science University. We prepared a letter telling of Brent's diagnosis and describing the disorder affecting him. We sent it to all of our friends and co-workers, appealing for their help and asking them to pass it along to anyone they thought might help.

We also reminded them that most companies have a charitable donation matching plan. And we told them Gaetano's company would match 100% of his contributions, up to a limit. (to a maximum of \$1000 for the charitable agencies and \$5,000 for each University up to \$20,000 per year per employee.)

Some friends and relatives had their donation matched at a 200% rate by their companies. Others opted for payroll deductions that would trickle in throughout the year along with their employer's match.

We received response beyond our expectations. The donations to both the NBIA Disorders Association and to Dr. Hayflick's research group exceeded \$20,000. More than \$10,000 of that came from companies' matching gift programs.

We could not hold back our tears at the overwhelming show of support by our dear friends and a few good people we did not know.

We encourage all of you to try this as a method to raise funds. It's a simple strategy but very powerful.

Please contact info@NBIAdisorders if you are interested in having a fundraiser. We have handouts and information on fundraising and will be happy to assist you with your planning.

A date with Senator Ted Kennedy's office

By Kris McGourthy

A trip to Washington, D.C., to attend a workshop turned out to be an ideal time to drop in on my U.S. senator, Ted Kennedy. Kennedy is sponsoring legislation that bans human cloning but protects stem cell research, something I strongly support. I wanted Kennedy's office to know of my feelings, and I wanted to offer to help get the bill passed, if they could use me.



Matt, 6, Michael, 15, Nicholas, 9 months, and Bobby, 11. The McGourthy children are from Middleboro, Massachusetts and the two oldest boys have PKAN, a form of NBIA.

Members of Congress are always looking for "real people" to testify about how their lives are affected by legislation.

My visit was in February, when NBIA Disorders Association President Patty Wood and I were in Washington to attend a workshop and to learn more about the bio bank, which our association is joining as a founding member. I called ahead to Kennedy's office and made an appointment to chat about the stem cell legislation, which has been stalled in Congress.

I prepared a letter of introduction, including pictures of my boys, articles that had been written about them, a brochure on NBIA, along with the organization's newsletter. I explained that stem cell research was important because two of my sons have the disease, and a new child born last year is a carrier. A fourth child is not affected. The stem cells from my youngest were preserved at birth and are now being stored for possible future use.

U.S. House and Senate members often travel to their home districts to meet with constituents, either at public meetings or in

one-on-one sessions. In-district meetings are an option for those of you who can't travel to Washington but want to bend your representative's ear. In Washington, visitors don't always get to meet with the member of Congress; often a key staff member is delegated to meet with constituents.

Patty and I met with a legislative fellow, Jennifer Leib, who works on health legislation. As it turned out, she was very familiar with NBIA. She had volunteered at our first family conference in 2000 as a graduate student at Johns Hopkins University! She was very helpful in explaining how things work in getting a bill through Congress and how we could check its status on the Internet.

The stem cell bill was referred to the Judiciary Committee a year ago and could die there, but I let Jennifer know I'd be willing to testify at a Senate hearing on its importance if they are looking for someone.

I would encourage each of you to consider introducing yourselves to your U.S. representatives and senators. Let them know, either through personal visits or letters, how you feel about issues that concern you. The more they hear from the rare disease community about our needs, the more opportunity they have to understand, learn and, I hope, be more responsive to our needs.

If you would like to know who your representatives are and how to contact them, visit the House of Representatives at: <http://www.house.gov/writerep> and your Senators at: http://www.senate.gov/general/contact_information/senators_cfm.cfm

NBIA Brochure Available

We now have brochures available with updated information on NBIA. These trifold are perfect for fundraisers or to give to friends and possible donors for information on our organization and the disease.

Perhaps you would like to write letters to family and friends as the Bonfiglios did (see story on pg. 2) and include a brochure with your employee contribution solicitation letters. Or maybe you are planning a fundraiser and want to have them as part of your information packet.

Please contact us at info@NBIAdisorders.org or (619) 588-2315 if you would like a free supply.

Lamos

(continued from pg. 1)



Barbara Belcher (left) at Vocational Training.

deficit disorder but no hyperactivity. She began to walk more awkwardly, just like her sister had. All three of my children were wearing out the toes of their shoes and sneakers because they were not walking correctly. MRIs showed iron deposits on the brain. Each was diagnosed as a teenager with NBIA.

My fourth child, Ben, 19, also had an MRI. It showed no iron deposits. He does not have NBIA like his siblings and spends most of his time with his father. We are divorced, and the primary responsibility for caring for the three affected children is mine.

I work full-time as a nurse and have been one since 1976. I had never heard of Hallervorden-Spatz (former name of NBIA) and would have been much more cautious about having children had I known this could happen. What little I found to read at the time was not encouraging: an end prognosis of dementia, muscle weakness and paralysis. I really had a hard time believing the diagnosis or I probably would have found this support group sooner. I was in a state of denial for about five years. But it gradually became real to me.

Luckily, I found a most wonderful neurologist who had dealt with NBIA previously. The neurologist was into research and genetics and put me on to the NBIA Disorders Association where I found much needed support and information.

We sent Becky's blood to a Chicago lab and she did not have the *PANK2* gene. I think this goes hand in hand with them not seeing the eye of the tiger on the MRI, so I was not surprised. Even though Bruce and Barb also have NBIA, Becky's case is much worse. We don't know why.

After the divorce, I've tried to juggle my children's care by taking one day at a time and keeping a positive outlook. I believe everything happens for a reason. We are constantly adapting, making necessary changes in our home to accommodate a

wheelchair or a walker. I am always amazed at how positive the kids remain, and that inspires me and helps me cope.

And the good news is, their stories don't end with the NBIA diagnosis.

Bruce graduated from high school in 1998. He went on to community college, until we figured out it wasn't for him. He got his driver's license at 18, and loves to drive. Vocational & Educational Services for Individuals with Disabilities helped place Bruce in a job as a mail clerk at the state education building in Albany. They provided a job coach, and Bruce proved to be a hard worker. He is well liked by his co-workers.

In July 2002, Bruce got a permanent state position. He is fond of saying, "I will not let this disability keep me down, I am going to beat it." Bruce is on Artane for dystonia and Baclofen for spasticity. He has been getting Botox injections in his legs for a couple years.



Bruce Belcher (right) with a friend from bowling.

Recently, his balance has worsened, and we are considering a Baclofen pump. He had a test dose inserted with a lumbar puncture and responded well. Bruce bowls once a week in a league with his sister Barb. He enjoys computer games & football.

Barbara graduated high school in 1999. She always struggled with her schoolwork but tried her best. Barb did some vocational training and Vocational & Educational Services for Individuals with Disabilities worked with her in a few job situations with a job coach. It didn't go well for her. We finally got her in a sheltered workshop where she stuffs envelopes and folds papers. Barb is very happy there.

(see Lamos on pg. 5)

Lamos

(continued from pg. 4)

Once a week, she gets physical therapy and attends group therapy where she gets help dealing with relationships and develops coping skills. She loves music and computer games and is very outgoing and friendly. She loves to talk and greatly enjoys the bowling league.

Like her brother, she also takes Artane and Baclofen. Barb has worn ankle/foot orthotics, which are leg splints that help her walk better, for the last two years. She also has been getting Botox injections for the last 1-1/2 years in her legs. She uses a walker when she goes to the mall or when walking long distances.

Since October 2000, Becky has slowly lost the ability to care for herself; she has not been able to talk for about two years. Since January 2003, she hasn't been able to walk.

Becky is the hardest for me to talk about. She graduated from high school in 1995 and was an excellent student. She was always a happy, talkative child and teenager who made friends easily. She enjoyed listening to the Beatles music and collecting trivia about them.

When we first got her diagnosis after investigating Becky's clumsiness, I just couldn't believe this bubbly, vivacious young woman would not have a promising future. She went to community college part time with help from Vocational & Educational Services for Individuals with Disabilities. While working toward an associate's degree in human services, NBIA started to take its firm grip on her, making learning — and even walking — more difficult. She had earned 24 credits, and wanted to work helping others with disabilities as she was just beginning to understand her own disability.

Since October 2000, Becky has slowly lost the ability to care for herself; she has not been able to talk for about two years. Since January 2003, she hasn't been able to walk. Becky has also been going to a day program since October 2000, it was at that time I realized she was not safe to care for herself home alone. It is open Monday - Friday from 8 am to 4 pm. She could still walk and talk when she started and they have experienced with me her steady decline, feeling our pain. She has been on Eldepryl, which did help her walk a bit better for a short time, but now she is just on Artane and Baclofen. Recently, we added Klonopin because she was starting to have involuntary muscle spasms that were quite painful.



*Becky Belcher's high school graduation in 1995.
With her is her great grandmother.*

The Klonopin has helped but makes her sleepy. I can get her to smile for me very easily when I sing along with a Beatles song; she finds that quite funny.

Ben, my youngest, is a college freshman. He is quiet and sensitive. He sometimes feels guilty for not being affected by NBIA and wonders why. He enjoys playing guitar and is part of a heavy metal rock band. He tells me that is how he gets out his frustrations.

My life is busy and full and often not my own. Yet this is MY life. I hope and pray for a medical breakthrough that might stop NBIA from robbing our children of their lives. Actually, the thing I have the hardest time doing is telling others about my children and accepting help when I need it. I think it's partly a mother/nurse thing, the feeling that I can do it myself.

I don't like to ask for help, but I have come to admit that I can't do it all myself. My sister is about 30 miles away, and I have a few close friends who are major sources of support. For the last two years, I have an evening aide from 2 p.m. to 8 p.m., Monday through Friday, who has been invaluable to me and who has made my life much easier. She is only paid to care for Becky but very willingly helps with Bruce and Barb. For the last six months she also works one weekend day.

Being part of the NBIA support network is one of the best things I have done for myself. I know I am not alone. Yes, I get exhausted. But, could I not do what I do? No, these are my kids. There is really no choice.

I think the most important thing I have learned living with my three adult children with NBIA is not to take anything for granted. Take each day for what it is, and enjoy that day.

Bank

(continued from pg. 1)

in how the disease is described in the medical literature. Also, researchers don't typically follow up on affected individuals after they donate, so the progression of symptoms is not seen and valuable information is lost.

Many times, affected individuals and their families never hear back about any research findings on tissue donations. And some researchers are reluctant to share their samples with colleagues or may have legal problems doing so because of regulations.

With a bio bank, researchers given access to samples will report back to the NBIA Disorders Association on their results, and the organization will inform families.

If the bio bank lives up to its potential, it can change the way research is being done today.

There are a variety of advantages for those who participate. Privacy and confidentiality are assured for those donating blood and tissue samples. Only samples stripped of identifying information are given to researchers. Affected individuals and their families only have to donate once, rather than multiple times for different research projects. Donors go through a consent process to help them make the decision freely before participating.

Researchers have access to samples that follow the same collection and storage protocols and access to follow-up data. A goal is to have a research database that will help researchers and pharmaceutical companies study similar symptoms across different diseases, possibly opening the door to new treatments.

Dr. Susan Hayflick, a key NBIA researcher, said she strongly supports the decision to join the bio bank.

"A significant hurdle to attracting new researchers to study a rare disorder is limited access to tissues," Hayflick said. "Most scientists are not physicians and so do not have a patient connection for obtaining tissue for their studies. Just as fundraising has encouraged and supported new NBIA investigators, better access to tissue will remove a key barrier to progress in understanding and treating the NBIA disorders. Participation in the bio bank is a very wise way for the NBIA Disorders Association to support research."

The Genetic Alliance Bio Bank is still in its infancy. It was incorporated on Oct. 14, 2003 in Delaware with Sharon Terry as president, husband Patrick Terry as secretary and Joan Scott as treasurer.

Sharon Terry is also president and chief executive officer of the Genetic Alliance, an organization that helps individuals and

families living with genetic conditions. She is the founding CEO of PXE International, a non-profit dedicated to finding a cure for pseudoxanthoma elasticum. She's on numerous advisory boards at the National Institutes of Health, Johns Hopkins University and the Centers for Disease Control and Prevention, among others.

Patrick Terry is the president of the International Genetic Alliance and the co-founder of Genomic Health, Inc., a business dedicated to improving the quality of treatment decisions for patients with cancer. Joan Scott is the Deputy Director of the Genetics and Public Policy Center in Washington DC.

The bank's estimated budget for 2004 is \$97,182. Seven organizations, including ours, have joined and agreed to pay entry fees of \$13,883 each as founding members. Annual fees in future years will either be \$5,000 or \$10,000, depending on how many groups join.



Founding members of the Genetic Alliance Bio Bank meet for a workshop Feb. 14-15 in Washington DC.

Change of Address

Please notify us if your address changes. We send our newsletter out non-profit bulk mail, and they do not forward mail under that rate, even if you have filed a change of address with the post office.

The only way we will know where to send your newsletter is if you tell us — so please remember to do this in the future if you move. You can send an e-mail to info@NBIAdisorders.org or notify us via postal mail. Thank you.

Manage your own medical journey

What can consumers of genetic services do to optimize the experience of living with a genetic condition and ensure they are receiving the best possible health care? Patricia Foote, author of *How are you? Manage Your Own Medical Journey*, shares her research on this topic. Patricia lives with von Hippel-Lindau disease.

Patient Strategies for Good Outcomes

Become the team leader of your medical team. Remember that *you* are paying, so hire professionals who are competent, compassionate and communicate well.

Understand your condition. Today, with the use of trustworthy sites on the internet such as the National Institutes of Health, the National Library of Medicine, the Office of Rare Diseases and the Genetic Alliance portal to more than 600 resource organizations, it is becoming easier to find quality information. Lay advocacy groups are becoming experts in their specific condition. This enables consumers to prepare for doctor visits with a list of intelligent questions and to have information about different treatment options, including clinical trials.

Manage the routines of dealing with your condition. Remember to take medications and attend medical appointments. Keep a health history form up-to-date so that this can be easily sent to new doctors and also for your own ease in remembering what screenings, medications, etc., you have had. Monitor diet, rest and exercise.

Acknowledge the emotions that come with the diagnosis of, or life with, a genetic condition. Recognize fear, anger, depression, frustration, and isolation. Seek professional help if things are really bad or perhaps locate a support group for people with your condition. Find ways to reduce stress that are enjoyable such as walking, reading, listening to music. Be sure to schedule a break for yourself if you are the primary caregiver.

Reach out to friends and family, even if activities have to be modified. Try to maintain relationships by clear communication: "I need," "I feel," "I'm afraid of." Be specific when asking for help: "Could you please go grocery shopping for me on Thursday?" "I'd love it if you could prepare dinner on Thursday," "Will you please read the newspaper to me?" "Will you just sit next to me, hold my hand and say nothing?"

(see *Strategies* on pg. 8)

Tackling NBIA pain with morphine: Difficult issue requires much thought

By Dianne Gray

Watching a loved one with NBIA in pain is very difficult, especially when the exact source of the pain isn't known because the person can't tell you. Knowing how best to treat that pain can be even harder.

My son, Austin, is now 13, but more than three years ago, physicians believed that we were "at the end of the road" and that Austin was certainly near death. During the summer of 2000, he was in tremendous pain from dystonia in his back. Our doctors also said he had extreme bouts of inspiratory stridor, (difficulty breathing due to tightening of the muscles in the throat area), as a result of dystonia in his vocal cords. At times, he felt like he was suffocating.

After nearly a month in the hospital, the pain management team consulted with us and advised us that morphine was the only way to relieve his pain. We had certainly tried everything else at our disposal. The physicians suggested a morphine drip.



Austin Gray, 13, from Naples, Florida.

(see *Morphine* on pg. 9)

Strategies

(continued from pg. 7)

Stay apprised of health insurance issues. For example, what procedures require pre-approval; what is your annual deductible; what is your lifetime cap; what protections do HIPPA and the new HHS Privacy Regulations offer?

Stay sensitive to privacy issues. Who needs to know the information? If in doubt, don't disclose. Never give blanket disclosure authorization.

Maintain a positive outlook – an “I can do” attitude. Replace negative thoughts with positive action – not “why me?” but “what can I do?”

Designate someone to coordinate your medical management for you if you are unable to take on a proactive role.

Learn to accept your diagnosis and live your new life. Integrate the business of living with a chronic condition into your life and move on. Remember, you are much more than your disorder. Accept and understand what is “normal” for you, which may include an increased amount of time spent on medical issues and some physical discomfort. It may not always be possible to “win” the fight with a genetic condition – some are terminal. The knowledge gained from the battle could be considered precious, and victory a deeper love of those who shared the journey.

Realize that sometimes blessings and insights come from living with a genetic condition. Perhaps you learn to set priorities, value the use of time, become more empathetic, learn who your true friends are, and take care of unfinished business.

Think of all of the above in simplified terms: think of the five vowels – those little letters that all words need:

- A Attitude
- E Emotions
- I Information
- O Outcomes
- U Understanding

Maximizing the promises of medicine. A PBS TV special, *Critical Condition*, stated the following: “The quality of your healthcare depends as much on you as on your doctor, on your being informed, on your asking the right questions, on your being your own best advocate. Your life may hang in the balance.”

To reap the promises of medicine in the 21st Century, we must work proactively to manage our healthcare and live our lives to the fullest.



Andy Blyler
July 26, 1991 – Nov. 16, 2003

Some people come into our lives and quickly go, others stay for a while and leave footprints on our hearts and we are never, ever the same.

Andy loved going to school. He attended Quail Run Elementary where Mrs. Holt, Myrna and Teresa were his teachers, and Katie was his girlfriend. He rode bus 106. Jim and Hal, the bus driver and attendant, were his surrogate grandpas. Hal called Andy “Hollywood.”

Chris Gran was Andy's caregiver. She also was his friend, and will always be considered part of our family.

His nickname at home was “Rooney.” His favorite toys were his penguin he named Martin, and bunny rabbit. He loved the Disney channel, music, animals and the color green. I always said that if he could pick a career, he would work at Sea World with the dolphins or penguins. In 2002, Make-a-Wish fulfilled his dream of petting a penguin with a trip to Orlando, Florida. It was an awesome week and we will always treasure the memories.

Andy was not only our son but also our teacher and our best friend. He touched many people in his short life. He had an infectious smile and a personality to match. He taught us unconditional love and what truly matters in life. From him we learned this: Don't take the time you have with your children for granted. Don't pass up the chance to hold them, to tell them you love them. Make the most of every second.

Andy left a huge hole that will never be filled. But he forever changed our lives in just the short time he was with us. Thank you, Andy.

Love,
Mom, Dad and Christine

Morphine

(continued from pg. 7)

A local hospice organization has been managing Austin's pain with morphine ever since. Baclofen, phenobarbital and valium are still part of his medical regimen, but I believe morphine has kept him from suffering the extreme pain that he had experienced before we began this treatment. As an added benefit, Austin has gained about 30 pounds and grown in height because of his increased comfort level.

We also have added normal saline treatments and Atrovent to his respiratory regimen to alleviate breathing difficulties. When necessary, hospice recommends a low dose of Roxanol be added to his breathing treatment to prevent the feeling that he is suffocating.

While we are not sure if Austin will ever be morphine-free, we are thankful for the many days that he has been fairly comfortable.

Typically, when NBIA patients suffer from pain, caregivers contact primary care providers, often the patient's neurologist. Although the doctor may feel that morphine or Roxanol may be necessary, the physician might not feel comfortable dosing to the level necessary to reach pain-free status.

At that point, a pain management specialist or a hospice program are options. Frequently, pain management specialists connected with children's hospitals are familiar with narcotic levels and dosage amounts for pediatric patients and patients of rare disease. Contacting hospice does not mean that caregivers are preparing their loved ones to die. It simply means they are accessing an important resource to provide comfort.

It is important that before morphine is used the patient "receive an exhaustive evaluation for a treatable cause of the pain," said Dr. Susan Hayflick, geneticist and leading expert in NBIA at the Oregon Health & Science University. "Too often these are missed (a broken bone, bleeding stomach) and just the pain is treated."

If an examination fails to reveal the cause of the pain, caregivers or patients must ask more difficult questions. Has every diagnostic tool been used and are we ready to treat the pain in the absence of understanding the cause? If the caregiver were in pain, would he or she not turn to the other person in the room and demand some sort of pain medication? Do the patients with NBIA deserve any less?

"Worsening pain does not indicate that the person is nearing death," Hayflick said. "Pain episodes seem to come and go for most

patients. Sometimes there is a treatable reason and sometimes not. I think there is little, if any, correlation between worsening pain and worsening disease."

Many caregivers wince at the thought of morphine. They might associate the drug with end-of-life care or worry whether there will be anything stronger if needed later. Will the morphine hasten their loved one's death?

Dr. Deidre Woods, medical director of Hospice of Naples, Fla., said morphine is not necessarily reserved for end of life nor does it bring on death.

"In fact, patients frequently express a new lease on life and are able to eat or sleep comfortably now that they are pain free."

"In fact, patients frequently express a new lease on life and are able to eat or sleep comfortably now that they are pain free," she said. "Roxanol use in nebulizer treatments also can help a patient to breathe much easier especially when the patient suffers from the debilitating feeling of suffocating due to inspiratory strider. This brings a great deal of comfort to a patient and they can lead more productive and fulfilled lives with morphine as part of the medical treatment program.... If the patient requires less medication, we can decrease the amount of morphine until the patient is comfortable."

Woods said that respiratory problems occur mainly in rare cases where extraordinary high doses are given, especially if the patient has never had these types of drugs before. It is important to start at very low doses, then increase the medication level as needed.

The use of morphine in NBIA patients is a subject of debate and will continue to be a topic of consideration as long as our patients continue to suffer from intense bouts of pain. While we hope for a cure, many of us will continue to seek out information on pain management. Morphine may not be the only answer, but for some NBIA patients, it is an important part of a regimen of medications that attempt to keep them pain-free.

Web site photos

We would like to update and add more pictures to our NBIA families page. If you have previously sent in a picture and would like to send in a more recent one, please send the new photo (up to two per family are permitted) as an attachment via e-mail to info@NBIAdisorders.org or a hard copy via postal mail. NBIA families who have not been represented are invited to add their families to this portion of the Web site.

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

Sabrina & Alyssa Barbiero
Rosendale Elementary School Main Office Staff

Brent Bonfiglio
Catherine Amouris
Sean Dirzadeh
Ann Lencioni
Nancy Lueder Miora
Alison Mochizuki
Mr. & Mrs. Gene Mochizuki
Mr. & Mrs. Daniel Perez
Ann Marie Sheehan
World Reach, Inc.

Christy Davis
Peggy Davis

April & Tracie Flinn
Vera Ross
Sue Spradley

Becky Lamos
Marion Hermann

McGourthy Family
Judith Elliott
Paul & Mary Tierney

Sondra Roberts
Jane Roberts

Julie Smyth
Roy Smyth

Kimberly Wood
Georgetta Gambetta
Regina Neal

In Memory Of

Leonard Bononomi
Mr. & Mrs. Fred Knodle

Andy Byler
Black Canyon City Riders
Patty Carney
Carol Hines
Linda Jones
Kim McMillan
Terri Moore
April Munson
Mr. & Mrs. Ronald Paris
Ed Ramold
Mr. & Mrs. George Reichert
Ann Vaninetti

Carmen & Mary DelGrossa
Angela Steinker

Wendy Devens
Lee Demellier
Dennis & Judith Devens
Peter Latona
Mr. & Mrs. James O'Hara
Maria Valentino

Clarence Roser
Anella Roser

Jacob Wylie
Apache Truck Lines
Ida B. Lusk Circle
Janice Jones
Rhonda Jones
Robert & Julie Larue
Jonica S. Lasiter
Elise & Norris McDivitt
Mr. & Mrs. Mike McGourthy
Evelyn Mizer
Mike & Valerie Smith
Dorothy Walk
Jo Ann Walk

Richard H. Tapke, MD
Mr. & Mrs. Tom Tapke

Tony & Jean Venuto
Mr. & Mrs. Tom Tapke

Marguerite E. Weiseth
Chris Cleath
Valerie DeGolier
Dan & Mary Lou Haffner
Mark & Tami McGruder
Tom & Val McGruder
Dave & Vicky Terwilliger
Dan & Pat Willette

You know you have a child with special needs when:

- You compare Emergency Rooms instead of grocery stores.
- You compare your child's oxygen saturations.
- You teach your child HOW to pull things out of the cupboard, off the bookcases, and that feeding the dog from the table is fun.
- Everything is an educational opportunity instead of just having plain old fun.
- You cheer instead of scold when your child blows bubbles in their juice while sitting at the dinner table (that's speech therapy), smears ketchup all over their high chair (that's OT), or throw their toys (that's PT).
- You fired at least 3 physicians and can teach your family doctor a thing or two.
- You have been told you are "in denial" by at least 3 medical or therapy professionals. This makes you laugh!
- You have that incredible sinking feeling that you've forgotten SOMETHING on those few days that you don't have some sort of appointment somewhere!
- You get irritated when friends with healthy kids complain about ONE sleepless night when their child is ill!
- You keep an appointment at the specialist even though a tropical storm is raging because you just want to get this one over with and you waited 8 months to get it and besides, no one else will be there!
- When the Doctors/Specialists/Hospitals etc. all know you by your name without referring to your chart.
- You pureed turkey on Thanksgiving.
- Your child's medical file is four inches thick and growing.
- You have a new belief that angels live with us on earth.
- You don't take a new day for granted.

Borrowed from: Mothers from Hell, Brimstone Bulletin (Spring 2003)

MESSAGE FROM THE PRESIDENT

Four days in Washington



Patty Wood

Valentine's Day weekend was special for me this year. Not because I spent it in the usual way we think about (darn!) — but because I felt like I was on the threshold of something big, something that I think will be important to all of our futures.

It was a two-day workshop in Washington, D.C., on the Genetic Alliance Bio Bank for the founding members. It laid the groundwork for what the bio bank will do to advance the study of NBIA and other rare diseases. All of those involved share a common hope: that the bio bank will lead to new treatments and possibly a cure for the diseases being studied.

I am grateful to the Wright Foundation for giving us the opportunity to be on the cutting edge of this exciting endeavor.

Yes, being part of the bio bank will definitely require a big commitment on the part of our organization and NBIA families. But the potential benefits make it very worthwhile. To learn more about it, please read the story about this on page one and stay tuned to learn how you can participate.

While in Washington for the bio bank workshop, I also attended the National Institute of Neurological Disorders & Stroke Advisory Council meeting. This council meets three times a year for two days each time. Along with the usual business, there were two scientific presentations, one on strokes and another on a promising new treatment for Parkinson's disease. The Parkinson's presentation was especially interesting in light of some of the similarities between that disease and NBIA.

Attending the council meeting was Dan Tagle, program director for neurogenetics at NINDS. He is responsible for helping facilitate new research for NBIA. We had lunch together to discuss our planned Scientific Workshop on NBIA, tentatively scheduled for Spring, 2005.

We will be bringing in 20 to 30 researchers from around the world to discuss NBIA research and setting research priorities for the future. Our plan is to hold our third family conference in conjunction with the scientific workshop, just like we did in 2000 at our first family conference in Bethesda, Md. We are again hoping that families will be able to meet with researchers and ask questions.

Also while I was in Washington in February, board member Kris McGourthy and I scouted out a possible hotel site for the 2005 conference. Planning for these events requires a lot of advance work.

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.

- | | |
|-------------------------------|----------------------------|
| Bert & Shirley Babb | Mr. & Mrs. John McMahon |
| Brian & Karen Baker | Dorilee Male |
| Brian & Carol Barnes | Maxtor Corporation |
| Becker Construction | David & Mary Miller |
| Chase Adams Designs | Lori Mitchell |
| Angela Chicco | Priscilla Muscolino |
| Mr. & Mrs. J.V. D'Ambrisi | Irma Navarrette |
| Mr. & Mrs. Lawrence Delamater | Niskayuna HS Key Club |
| Diego North America | Nancy O'Donnell |
| Foundation | Douglas Ornstein |
| Christine Feeney | Jerry Oshiyama |
| Mr. & Mrs. Peter Flanagan | Mr. & Mrs. David Rich |
| Nadia Gallo | Anna Riley |
| Kathy Giebel | Patricia Rodriguez |
| Mr. & Mrs. David Gorman | Mary Ann Roser |
| Charles & Judith Griswold | John Ryan, Jr. |
| Guidarelli Contracting, Inc. | Sacred Heart Church |
| Mr. & Mrs. Homer Harding | June Schechter |
| Mr. & Mrs. George Jones | Jane Sebeck |
| Mr. & Mrs. Lowell Judd | Veronica Serviss |
| William Kelso | Mr. & Mrs. Jack Smyth |
| Mary E. King | Maxine Smyth |
| Angelika Klucken | Mr. & Mrs. Robert Starling |
| Joanne Krchniak | Cynthia Taillon |
| Ella K. Laird | Ellen Tolley |
| Cheryl Lamos | Mr. & Mrs. Dan Venuto |
| Lee Hamer Memorial Charities | Scott & Judith Wakefield |
| Mr. & Mrs. John McMahon | Mr. & Mrs. Rod Walker |

We even worked in a little lobbying. We called on Kris' U.S. senator, Ted Kennedy. Kris is very interested in stem cell research, and wanted Kennedy, who is sponsoring legislation on stem cell research, to know she'd be willing to testify in favor of his bill, which has been languishing in a Senate committee for more than a year. (Please see Kris' story on pg. 3.)

As you can see, we packed a lot into four days, but you will be hearing about the work we started for a long time to come.



Patty Wood at Union Terminal in Washington DC in February.



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

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