Men, women and children came from across the United States and from Australia, Canada, the UK, Norway, Mexico, France, Chile and the Netherlands to our Tenth International Family Conference, held May 30 to June 2 in Charleston, S.C.

In all, the conference attracted 77 families — half of them first-timers — for a total of 285 participants. Fifty-seven of them were NBIA-affected individuals. In comparison, our first family conference, held in Bethesda, Md., in 2000, drew 18 families and 70 total participants.

My, how we’ve grown!

Because of the ever-increasing number of NBIA families attending, we switched from pre-conference personal family appointments with our NBIA experts to clinical sessions for BPAN, MPAN, PKAN and PLAN. These sessions were open to all families, instead of only first-timers, as we’ve done for the past few conferences. Families were asked to submit questions for the clinicians beforehand and these were answered during the sessions, along with the

Clinical trial of Retrophin drug fails; shows no benefit for PKAN patients

A much-anticipated drug therapy has failed to show any benefit for individuals affected with PKAN, or Pantothenate Kinase-Associated Neurodegeneration, one of the most common forms of NBIA.

The drug’s maker, Retrophin Inc., announced the disappointing results Aug. 22 for its Phase 3 Fosmetpantotenate Replacement Therapy, or FORT, study.
What is NBIA?

Neurodegeneration with Brain Iron Accumulation (NBIA) is a group of rare, genetic, neurological disorders.

The common feature among all individuals with NBIA is iron accumulation in the brain, along with a progressive movement disorder. Patients can plateau for long periods of time and then rapidly deteriorate. The most common symptom is involuntary muscle cramping, called dystonia.

Symptoms vary greatly from one person to the next, partly because the gene affecting them can differ. Different mutations within a gene also can cause a more or less severe form of the disease.

The movement disorders can result in clumsiness, difficulty controlling the body and speech problems. Also common is a degeneration of the retina, which causes night blindness and a loss of peripheral vision.

Some individuals eventually lose the ability to walk, talk or chew food and become totally dependent on others for all their needs.
Conference
(continued from pg. 1)

experts sharing other clinical information.

Also for the first time this year, we invited U.S. clinicians who have seen several NBIA individuals to participate in our clinical sessions and receive hands-on training to examine individuals representing various NBIA disorders. Five attended and called it a wonderful opportunity to learn more about NBIA and be a part of our community. We plan to continue this program at future conferences.

This was the second family conference to include breakout sessions to give participants more choices. This year’s sessions allowed families dealing with the same NBIA disorder to break into groups to get research updates, share information and provide support to each other.

Other breakout sessions included an update on the completed deferiprone trial for PKAN, communication when speech isn’t possible and research steps for rare disorders. Single sessions for all included tips for creating a future care plan for your child, cannabis use in NBIA disorders and the development of PKAN therapeutics. (You'll find articles on many of these topics in this newsletter.)

The opening keynote address on Friday by Gina Perri Cannady of Jenkintown, Pa., inspired many participants and tied into the conference’s theme, a “Beacon of Hope.” (see article on pg. 6)

Our memorial event on Friday afternoon for those NBIA individuals no longer with us was a poignant tribute. With the meeting space darkened and gentle music playing, 92 volunteers carried glowing luminaries bearing the name of an individual who we’ve lost to NBIA. Each name was displayed on large screens near the stage.

Saturday afternoon we went to a local park for lunch and enjoyed spending time together in a relaxed atmosphere. We played games and enjoyed watching a competition between teams led by Drs. Susan Hayflick and Penny Hogarth eating donuts hanging from strings.

That evening we held another first — a thank you reception for our major donors, sponsors and fundraisers to recognize their contributions and hard work. Afterward, we held our dessert social and danced while a DJ played music.

Also new this year: We livestreamed and taped all of the general sessions and many of the breakout talks to accommodate those who couldn’t attend.

You can find a link for the 22 taped sessions at https://nbiadisorders.org/2019-conference, along with videos of our closing ceremony, a photo montage of pictures from the conference and pictures of NBIA individuals gathered before the conference.

As you can tell, the conference was packed with activities and sessions — all of which required meticulous planning. We are grateful to the Family Conference Committee, led by board member Susan Laupola, and our Development Committee, led by board member Matt Ritzman. The volunteers on these two committees worked hard to make this conference one of our best ever.

None of this would have been possible without our conference sponsors who donated $78,966 to help us defray increasingly expensive conference costs. Retrophin, Inc., was our premier
Conference
(continued from pg. 3)

sponsor for the fourth time, and we had two elite sponsors, ApoPharma, Inc., and CoA Therapeutics, Inc., along with 12 others.

With donations of $5,654 mainly from individuals, we were able to provide scholarships to eight families, which covered their conference registration and four nights at the hotel.

Additional revenue of $34,816 came from registrations, merchandise sales, and silent auction and raffle items contributed by our families.

Conference expenses of $130,871 exceeded revenues of $119,436 by $11,435. We will make up the difference with general fund monies earmarked for family support.

We’re already working on the 2021 conference. We’ll announce when a date and site are chosen.

Conference Sponsors
NBIA Disorders Association is extremely grateful for the wonderful response in our request for sponsors to help us finance the Tenth International NBIA Disorders Association Family Conference. With their help, we were able to provide a forum where individuals, families, caregivers, educational and professionals joined to share experiences, learn from experts, and discover new resources.

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Colleen & Allan Lukoff
Regina Neal & Mark Botten

Conference Scholarship Contributors
NBIA Disorders Association would like to thank the following individuals, for it is through their generosity that we were able to offer scholarships for families and individuals who otherwise would have been unable to participate in the Tenth International NBIA Disorders Association Family Conference.

Lisa Aguirre
Jeff Berwick & Lai Kwok
Gaetano & Veronica Bonfiglio
Joseph Brostek
Marsha Bryan
Doug & Kimberly Burke
Bonnie Conners
Amanda Craft
Rosalind DiBiasi
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Megan Thomas
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David & Susan Tink
Mila Trnka
Seventy-eight PKAN individuals had completed the 24-week randomized, double-blind study, meaning that neither the patients nor the doctors knew who was randomly selected to get the drug or the placebo. At the end of the study, 76 patients decided to participate in the open-label program in which all received the drug.

Although the drug, Fosmetpantotenate, was observed to be generally safe and well-tolerated, the study found that it did not meet its primary or secondary endpoints, or outcome measures.

First, the study found no differences between those who received the drug and those who got the placebo. That determination was based on the extent to which individuals improved over the 24-week trial, based on a scale that measures activities of daily living, such as walking, eating and dressing. Those measures were specifically adapted for PKAN individuals using Part II of the comprehensive and widely referenced Unified Parkinson's Disease Rating Scale.

Second, the study found no measurable change on the same scale’s Part III score, which evaluates motor function, including slowness, stiffness and balance.

No data suggested that a longer course of treatment would change the outcomes, nor were any differences seen between classic and later-onset PKAN individuals taking part in the trial.

“We are very disappointed in the topline results from the FORT Study, particularly because we have seen the devastating impact of PKAN on patients and their families, and a significant unmet need remains with no approved treatment option,” said Retrophin CEO Eric Dube, Ph.D. "We would like to thank the patients, their caregivers, study investigators and our employees, whose dedication made this study possible."

The study gathered a significant amount of data, which is still being analyzed. Retrophin plans to present its findings at scientific meetings in the fall. It also will publish the findings in a peer-reviewed journal. Company officials said they hope the data will help inform future clinical studies for treating PKAN.

A letter to the NBIA community from Eric Dube, CEO of Retrophin, Inc., is on our website home page at www.NBIAdisorders.org
Many NBIA parents could empathize with Gina Perry Cannady’s rare disease journey because they’ve traveled the same road: The nagging fear that something is wrong with their child. The numerous doctor visits that yield no answers. And, finally, the devastating diagnosis. But Cannady’s journey also is one of hope.

Cannady, who lives in Jenkintown, Pa., delivered the keynote address May 31 at the Tenth International NBIA Family Conference in Charleston, S.C., where the theme was “Beacon of Hope.” She described how she, her physician-husband, Steven, and their children have been on a long odyssey with spinal muscular atrophy, or SMA, a rare neuromuscular disorder that causes muscle wasting. It is the No. 1 genetic cause of infant death in the United States.

The Cannady’s second daughter, Emma, born in 2009, seemed delayed in reaching milestones in comparison to her older sister. Cannady noticed Emma fell a lot, and she knew something was wrong. But doctors kept telling her she was “worrying about nothing.”

When Emma was 3, the family finally got an answer; thanks to an astute physical therapist who urged the couple to visit a neurologist. Emma was diagnosed with SMA, and there was no treatment.

At the time, Cannady was 37 weeks pregnant with her third daughter, Ruby. The baby arrived happy and full of life, but when she was just 10 days old, the couple got the news: Ruby also has the gene mutation that causes SMA. When it showed up, it was moving much faster than it had with Emma.

Cannady tearfully described her fears for her girls’ lives and the dark days that followed. But hope was coming. It arrived in the form of a clinical trial for the drug Spinraza.

“They wanted Emma to be a guinea pig,” Cannady said. Doctors told the couple that they would have to remove cerebral spinal fluid from Emma’s brain and that while they were hopeful, death also was possible.

“The risks were absolutely terrifying,” Cannady said, adding that she was very reluctant to enroll Emma in the trial. But her husband insisted, saying he believed in the power of science to help their girls.

Within a week of receiving her first dose of the drug, Emma walked six city blocks without falling. “Emma was getting better, but Ruby was struggling,” Cannady said. They wanted Ruby to get the drug, too.

“I have seen unbelievable progress in just six short years,” she said, “Our Facebook page used to be filled with obituaries.” 
But now the postings describe what SMA children can do.

But the U.S. Food and Drug Administration planned to stop the drug after Emma’s second dose. The couple fought back and managed to get a private meeting with the FDA. “We asked them not to stop this incredible science,” Cannady said.

The FDA resumed the trial, and Emma continued to receive the drug. Ruby was allowed to get it after she turned 2.

On Dec. 23, 2016, the FDA approved Spinraza as the first drug to treat SMA. Both girls take it, and while they are in wheelchairs and remain affected by SMA, they are doing well, Cannady said.

“I have seen unbelievable progress in just six short years,” she said. “Our Facebook page used to be filled with obituaries.” But now the postings describe what SMA children can do.

Days before the conference began, the FDA approved an innovative gene therapy for SMA. Emma and Ruby aren’t eligible for the therapy, Zolgensma, because patients must be younger than 2. But her girls are holding their own.

Cannady encouraged the NBIA audience to be patient, to not give up. She urged them to be advocates for their children and to dare to believe that better days are possible.

“Science takes time, but science gets there,” she said. “My girls play and explore because of that trial. Because of that trial, I get to watch them … be in this world. What the drug did for me is, it gave me permission to imagine my daughters’ future.”

Although Ruby is just 7, the family is making plans for her future: Celebrating her 10th birthday at the Eiffel Tower, no less.
No time like the present to plan for your loved one’s future care

Mary Anne Ehlert says growing up with a disabled sibling taught her the importance of planning for the future. As a child, her parents let her know they were counting on her to take care of her sister, Marcia, when they were gone.

As a working professional, Ehlert established a financial planning company in 1990 to focus on future care plans for a loved one with special needs. Sadly, Marcia, who had cerebral palsy, suffered a seizure in 1995 and died.

“Everybody in my family was devastated,” Ehlert told participants at the Tenth International NBIA Disorders Association Family Conference. “I couldn’t move on.” Finally, one of her mentors told her to “quit feeling sorry for yourself (and) use what your sister taught you.”

In 2003, Ehlert, a certified financial planner, established Protected Tomorrows, an Illinois-based company that offers families education to assist with care planning and access to advocates who can help, as well as resources and membership for professionals who assist families, plus other services.

“My mission in life … is to make sure our family member who has a disability has the best, the happiest and the fullest life they can have,” she said.

To that end, she outlined various steps family members can follow when developing a future care plan for a disabled loved one:

* Think about where and how the person will live if or when something happens to the caregiver. Consider what kind of living arrangement would make the loved one happy.

* Make sure that the schools the disabled person attends, the individual’s health care providers and health insurance companies have current information. Be sure to document everything; well-kept records are vital, not just for yourself but for others who may follow you.

* If you’re the primary caregiver, communicate with other family members about the disabled person so they can step in if something happens to you. Let them know how your home is set up, what the disabled person likes to do, whether they have a favorite chair or drinking cup and anything else that might be helpful.

* Become intimately familiar with state and federal benefits, such as Medicaid, Medicare, Social Security, Supplemental Security Income and other entitlements, that help cover costs involved in caring for your loved one. Learn the age at which the person becomes eligible for certain benefits and apply for them in a timely way.

* Because assets and income affect benefits, avoid giving financial gifts. Put the individual’s inheritance, if there is one, in a special needs trust (also called a supplemental needs trust). The trust covers everything that government benefits don’t cover, such as extraordinary medical care or vacation travel.

* Be sure to consult with an attorney to make sure you are doing things correctly. The Protected Tomorrows website lists lawyers who are versed in special needs trusts and other issues.

* If you are the primary caregiver, be sure to state in your will who will step in to care for the person if something happens to you. Communicate well with that person so he or she knows what to expect.

* Become familiar with ABLE accounts, a federal program that allows states to create tax-free savings accounts for people with disabilities.

About that money you raised: How is your board spending it?

By Rick Tifone

From bake sales to bike rides to golf tournaments, along with many other creative initiatives like virtual, online fundraisers, NBIA families and their friends have raised nearly $2.3 million for NBIA research since 2002.

So, how did the NBIA Disorders Association invest the dollars that families worked so hard to raise?

Many of you may know that the association puts the money into grants and contracts to advance NBIA research around the world. NBIA grants have funded basic scientific research and various projects; including work resulting in the discovery of some of the 10 genes now linked to specific NBIA disorders.

(see money on pg 8)
Money
(continued from pg. 7)

You may also know that donations of $1,000 or more can be designated for research into specific disorders. As the association's treasurer, I wondered how much we were investing in each disorder, so I dug a little deeper to follow the money trail since 2002. I reported on those findings at the Tenth International NBIA Family Conference in Charleston, S.C. You can find that recorded session online at https://nbiadisorders.org/2019-conference.

What I found was, since 2014 the largest share of the association's grant awards has gone to BPAN, Beta-propeller protein-associated neurodegeneration — the result of intensive fundraising by BPAN families. The BPAN gene wasn't discovered until 2012, more than a decade after the first NBIA gene was identified for PKAN. BPAN has received a total of $611,494 in research grants and contracts.

BPAN cases are accelerating. We are seeing about one or two BPAN families per week contacting us with a new diagnosis, and BPAN is quickly becoming one of the most common forms of NBIA. For a long time, it was believed that half of all NBIA cases were PKAN, or Pantothenate kinase-associated neurodegeneration. We now think that percentage is about 35% for both PKAN and BPAN.

The graphs on this page show by year and by disease, where the money has been invested. The category of NBIA is when research funded benefited all NBIA disorders.

How do we decide what to fund?

The association's board of trustees works with its Scientific & Medical Advisory Board to set research priorities, evaluate grant requests and hold researchers accountable for their performance. Researchers must meet certain milestones by specified deadlines, are limited to 15 percent in spending on overhead and must share information on their results to maximize the impact of every dollar.

When this organization was formed in 1996, very little was known about NBIA. Today, clinical trials are underway to treat PKAN, and researchers are laying the groundwork for future trials on other NBIA disorders.

For those of us with kids affected by NBIA, progress never seems fast enough. But when you consider how far we have come in two decades compared with the progress of better-known diseases, like Parkinson's, the record shows we've been efficient, effective stewards of your donations. Together, I believe, we are on the right course — the path to curing NBIA disorders.
Interest rising in cannabis for NBIA, but much more research is needed

Cannabis, commonly known as marijuana, is increasingly being used as a treatment for epilepsy and chemotherapy-induced nausea and pain. Now, caregivers and patients with movement disorders, such as dystonia and spasticity, are wondering whether it might ease their symptoms.

Some reports are encouraging, but there haven't been enough studies to conclude that cannabis will benefit NBIA individuals, said Dr. Jenny Wilson, an assistant professor of pediatric neurology at the Oregon Health & Science University and at Doernbecher Children's Hospital in Portland.

Wilson spoke at the recent NBIA Disorders Association Family Conference and described how chemicals derived from cannabis are being used for medical conditions. The main ones are THC, the psychoactive ingredient in marijuana that produces a high, and the more-benign CBD, which does not cause intoxication.

As of June, 33 states have legalized medical marijuana use, including 11 that also allow it for recreational use.

Although recreational THC use can have such side effects as reduced brain volume, depression and attention/memory problems (in adolescents), the adverse effects of CBD are "pretty mild," Wilson said. Those effects may include tiredness, diarrhea and reduced appetite.

Last year, the U.S. Food and Drug Administration approved the oral use of CBD-based Epidiolex for two rare and severe forms of epilepsy in patients ages 2 and older. "Overall, kids tend to tolerate it well," Wilson said.

Most of the data on cannabis for spasticity and dystonia stems from studies involving multiple sclerosis patients, Wilson said. A number of studies in adults who used cannabis containing CBD and THC orally or in spray form showed an improvement in their spasticity. In smaller studies, adults did not show improvement in dystonia, however.

In a study of 16 children and young adults with complex neurologic conditions and spasticity, 12 had improvements in spasticity after receiving drops of THC-containing cannabis for an average of 181 days. One patient reported vomiting and another experienced restlessness.

In an another study of 20 children with dystonia and/or spasticity who used CBD- and THC-containing cannabis, all patients had improvements in both conditions, as well as quality of life and sleep. Wilson said. There were side effects in some, however. Two had worsening seizures, two had behavioral changes and one had increased sleepiness.

As for cannabis use in NBIA, Wilson said she gets asked about it a lot. To date, no studies have been done, but the NBIA research group at OHSU conducted an international online survey of 44 PKAN patients on cannabis use, Wilson said. Eighteen responded, 11 of whom said they considered cannabis and seven of whom actually used it for dystonia, sleep, pain or anxiety. Several patients reported significant improvement in dystonia, but the results were mixed overall.

In general, cannabis products have the potential to improve some neurologic conditions, but more research is needed to ascertain whether cannabis is beneficial to NBIA patients. OHSU is considering surveying those with other NBIA disorders on cannabis use.

In the meantime, Wilson said she would not discourage parents from trying cannabis in a child who is suffering and hasn't been helped by other treatments. But, she added, "At this point, I can't answer, 'Should my child use it?'"
Mother shares ways to communicate when your family member can't speak

For a 22-year-old, MichaelAnn Byrne has notched an impressive list of accomplishments: jewelry designer, subject of numerous national news features, homecoming queen.

It's clear from listening to her mother, Sherry Byrne of Mechanicsburg, Pa., that she is proud of MichaelAnn. Byrne spoke at the Tenth International NBIA Disorders Association Family Conference about communicating with a loved one who is non-verbal. MichaelAnn has one of the more common NBIA disorders, BPAN, and can't speak or walk.

But what she can't do does not define her, Sherry Byrne said. "MichaelAnn is not her disability."

Byrne has worked with people who have development and intellectual disabilities since the early 1980s. She was trained to set goals for those individuals based on what they couldn't do. "Can you imagine working on the same goal year after year after year?" she asked.

In the early 1990s, she was introduced to "person-centered planning," which builds on individuals' strengths, wants and dreams. It presumes the person is competent and should be treated with dignity and respect, Byrne said. But how do you find out what's important to the person if they can't talk?

Bryne says most of communication is non-verbal. Where the person looks, their body posture and facial expressions are key. She can tell what MichaelAnn likes by her eye gaze, smile, and attention. MichaelAnn leans forward and brightens when she's engaged.

She uses an iPad with her daughter and discovered she was deeply interested in social media.

When MichaelAnn took an art class in the eleventh grade, she no longer came home from school and went straight to bed for a nap, Byrne said. She was excited and wanted to let her mother know what happened in class.

"Her art teacher was great," Byrne said. "She always asked MichaelAnn what she thought."

Byrne would help her daughter with her jewelry-making by asking her lots of questions while MichaelAnn was designing a piece: What colors did she want? What shapes? How big?

MichaelAnn made friends in that class, something that had been hard for her to do previously. It inspired her to ask the Make-a-Wish Foundation to help her gain 100 friends. Country music star Brad Paisley promoted her request in a video, and the students responded, asking her for lunch dates. Then, they chose her as homecoming queen. National news outlets, including the Today Show, Cosmopolitan and People magazine, picked up the story.

Byrne told the Huffington Post she hadn't realized how much it meant to her daughter to be homecoming queen. MichaelAnn's smiles and giggles that night, hours afterward, told her all she needed to know.

"It was amazing," she said. "I just have no words for that."
BPAN grant award takes a new approach to understanding the brain's iron problem

By Patricia Wood

A newly awarded grant to study BPAN is taking an innovative approach to investigating and potentially treating the problem of iron in the brain, a characteristic seen in NBIA individuals.

Dr. Lena F. Burbulla, an assistant professor at Northwestern University in Chicago, is the lead researcher on the grant award of 65,000 euros, equal to about $73,700. It is being awarded by the NBIA Disorders Association in collaboration with two sister organizations in Europe, AISNAF in Italy and Hoffnungsbaum e.V., in Germany.

Burbulla's proposal to study Beta Propellar-Associated Neurodegeneration, or BPAN, was the unanimous choice among five applications reviewed by two review groups, the Scientific Advisory Board and the Lay Review Board.

The study's objective is to define the mechanisms that cause BPAN to develop and progress, focusing on the role of iron. Burbulla's innovative hypothesis is that BPAN is caused not so much from iron accumulating in the brain but from a lack of "bioavailable" iron, which is needed for the cells to work properly. Bioavailability refers to the degree and rate at which iron is made available to the cell, its organelles and proteins. As an example, mitochondria — the "powerhouses" of the cell — need iron to generate the energy essential for a cell's survival.

To test her idea, Burbulla plans to do a series of experiments with the help of skin samples donated by BPAN individuals. The skin cells are used to generate stem cells, called induced pluripotent stem cells. Those cells are then reprogrammed in the laboratory to become brain neurons, in particular dopaminergic midbrain neurons — the cell type most affected in BPAN patient brains.

The study has the potential to lay the foundation for identifying targets for treating BPAN. It may also improve the understanding of the link between iron and neurodegeneration, while suggesting ways to treat other NBIA disorders.

Burbulla also will be testing new therapies using various technologies, such as three-dimensional tissue cultures of the brain that mimic in the laboratory what happens in BPAN individuals.

The study has the potential to lay the foundation for identifying targets for treating BPAN. It may also improve the understanding of the link between iron and neurodegeneration, while suggesting ways to treat other NBIA disorders.

Strong preliminary data with Burbulla as one of two lead authors has previously been published in the October 2018 issue of Brain suggesting that iron overload is accompanied by various malfunctions in the cells of the WDR45 gene, which is mutated in BPAN individuals.

Burbulla's 18-month study, starting this fall, is titled, "Mechanistic insights into iron accumulation in WDR45 mutant neurons linked to ß-propeller-associated neurodegeneration."

Her grant marks the second time our organization and AISNAF and Hoffnungsbaum e.V., have collaborated to award research grants. AISNAF will manage the grant, but all three organizations will have equal responsibilities and rights.
NBIA Disorders Association holds first Facebook Live event on gene therapy

By Patricia Wood

Gene therapy generated a lot of buzz at our family conference in May, with many participants wanting to know more about its potential to treat NBIA.

In response, the NBIA Disorders Association hosted its first-ever Facebook Live discussion on the topic Aug. 6. It featured two conference speakers, Lauriel Earley, a postdoctoral fellow from the University of North Carolina - Chapel Hill, and Susan Hayflick, a medical geneticist from the Oregon Health & Science University.

At the conference, Earley told families she has started working on gene therapy for PKAN, which our organization helped fund. On Facebook, she discussed the therapy’s potential for other NBIA disorders, focusing on BPAN.

She is using one of the most promising and attractive approaches to gene therapy, adeno-associated viruses, or AAV. The researcher makes this common, well-tolerated virus in a laboratory and removes the original genetic material in the virus, replacing it with a corrected copy of the human gene. It is then ready to be a delivery vehicle that holds the healthy gene inside.

When delivered, the healthy gene sits alongside the disease-causing gene, but unlike the disease gene, it can produce functional protein. She stated that since this method supplies a complete, healthy gene to the cells, the individual genetic mutation is usually not important.

Earley noted that, fortunately, all NBIA genes are small enough to be treated with gene therapy.

(see gene therapy on pg. 13)
Gene Therapy
(continued from pg. 12)

fit inside the virus and are good candidates for AAV therapy.

The blood-brain barrier can be crossed via lumbar puncture to deliver the AAV into the spinal fluid, (intrathecal delivery), which appears to be the best choice for treating BPAN because it reaches the whole brain region which is affected in this disorder. Ideally, a one-time treatment is all that would be needed.

AAV is popular due to its safety profile, and has been successfully used in clinical trials showing long-lasting improvements in the lives of many patients. The concept behind this treatment is simple, but can take many years of work to perfect for each disease.

AAV therapy doesn’t have room for regulatory information in the virus along with the healthy gene, so there can be problems if too much protein is produced that causes harm. This is something that isn’t known until you try it, so testing in large animals before using it in humans is important.

Virus production also must be tightly controlled to ensure purity and safety. Currently, only a few companies are capable of doing this work, causing researchers to book requests several years in advance. It’s also expensive.

"Because there are now two AAV gene therapies approved, there has recently been more interest in expanding the number of companies that can produce viruses using this method, but it will take a few years to bring everything on line," Earley said.

Earley stated that the path to a therapy consists of tests using patients’ cell cultures, which are readily available at the Hayflick lab; tests on small animal models (German researchers have a BPAN mouse model); safety tests on large animals; and clinical trials in humans.

Having strong natural history studies is critical. These studies, which show symptoms in an affected individual over time, determine if delivering a healthy gene has worked. This could avoid placebo-controlled, double-blind studies so that all participating patients would potentially benefit.

Because our researchers have established cell lines and mouse models, we have taken the necessary first steps. The next step is to reach out to scientists willing to dedicate several years to this work, provided we can help with start-up funds.

Earley has expressed interest in working on a gene therapy for BPAN.

"To my knowledge, there isn’t anyone currently working on an AAV based gene therapy for BPAN," she said. "The initial work of making the virus and testing it in the cells can take six months to a year. Testing it in mice can also take a year. For an estimated time line, I’ve heard about five to 15 years is reasonable once the work has begun. My understanding is that the current trial for GAN (giant axonal neuropathy) took about eight years from beginning the work to starting the trial."

Having strong natural history studies is critical. These studies, which show symptoms in an affected individual over time, determine if delivering a healthy gene has worked. This could avoid placebo-controlled, double-blind studies so that all participating patients would potentially benefit.

"The age of the patient does not, in principle, affect whether they can receive a therapy," Earley said. "People as young as one month old have been given an AAV gene therapy treatment. That said, the therapy will work best in cases where there hasn’t been significant disease progression because we can’t fix tissues that are already significantly damaged. Hence, we think in the case of neurological disorders, younger patients are going to be better candidates for gene therapy."

Hayflick said she thinks the BPAN brain shows damage even before birth, so there might still be developmental problems for BPAN individuals receiving gene therapy even if very young. But she feels gene therapy might help with stopping the progression of the degeneration in BPAN and the Parkinsonism features that occur later in the disease.

BPAN families sent in questions prior to the event and these were answered during the session which only lasted about 35 minutes due to some technical difficulties. Questions left in the comments feature of the video were also answered by Earley in the days following the live session.

Future sessions covering gene therapy for MPAN and FAHN are being planned, and will be announced on our Facebook page when a date is set.

You can view the recent session at https://www.facebook.com/pg/NBIADisorders/videos.
Deferiprone trial results produce positive findings for some with PKAN

The long-awaited results from the first international clinical trial for NBIA — testing deferiprone in individuals with PKAN — are in.

They show that the iron-chelating drug slowed the progression of the disorder in older patients with a later-onset, or atypical, form of PKAN, but did not have a similar benefit for younger patients with classic PKAN, which starts in early childhood.

In addition to those findings, the study showed that the drug successfully reduced the amount of accumulated iron in the brain for PKAN individuals, regardless of onset age.

PKAN, or Pantothenate Kinase-Associated Neurodegeneration, and all other NBIA disorders share iron accumulation in the globus pallidus structure of the brain. It remains unclear, however, whether excess iron causes NBIA or is brought on by some other problem.

The results of the trial, which was funded by a European Union grant titled Treat Iron-Related Childhood-Onset Neurodegeneration, or TIRCON, were presented at the Tenth International NBIA Family Conference held May 30 to June 2 in Charleston, S.C. The findings, which can be found on our website home page, were then published in the July issue of the medical journal, Lancet Neurology.

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The lead investigator of the trial in the United States, Dr. Elliott Vichinsky of the University of California, San Francisco Benioff Children’s Hospital in Oakland, discussed the results at the conference. He said that older and younger PKAN individuals showed improvement with deferiprone in dystonia of the lower face and lower legs, as well as in cognitive functioning, especially memory. But the benefit in the younger children, who tend to have a faster-moving, more severe form of PKAN, "wasn't statistically significant," he said.

The 18-month trial, which ran from 2012 to 2015, involved 88 patients from the United States, Germany, Italy and England. The trial met the gold standard for research. It was randomized, with some patients getting deferiprone orally and others getting a placebo. Afterward, the trial was extended another 18 months, and the drug was made available to everyone who took part in the trial.

"The drug was well-tolerated, and the safety profile was very good," Vichinsky told families at the conference.

But because the study did not meet a key goal — showing a statistically important improvement from deferiprone in all age groups — the U.S. Food and Drug Administration hasn’t yet approved it for PKAN. Consequently, Vichinsky encouraged interested families to contact the FDA to advocate for its approval.

To see his presentation, go to https://nbiadisorders.org/2019-conference where we have a link to all the sessions taped at the conference.

Longtime SMAB member stepping down

By Patricia Wood

After 14 years of service, biomedical company executive Steven Richieri of San Diego resigned in April as a member of our Scientific and Medical Advisory Board.

Citing work circumstances and personal commitments, Richieri said it was time to move on. "It has been both an honor and a source of great personal pride to serve on the SMAB," he said.

His departure leaves the board with six members who advise and assist the NBIA Disorders Association Board of Trustees with medical and research issues. The trustees plan to replace Richieri and increase SMAB membership in the near future.

He joined the SMAB in 2005 on the recommendation of then-
trustee Jeff Doerner. Our organization initially used the National Organization for Rare Disorders to review research grant applications and make awards for us. When we decided to take that work in-house, we wanted a diverse group of reviewers and asked Richieri to join us.

He has more than 35 years of experience in the health care industry and extensive experience with the U.S. Food and Drug Administration’s regulatory processes. Richieri is President and Chief Operating Officer at Banyan Biomarkers Inc., a San Diego start-up medical device company which recently received FDA clearance for the first in vitro diagnostic test to aid in the evaluation of traumatic brain injury.

We are very grateful to Richieri for his time, energy and commitment. We will miss working with him.

Recent NBIA fundraisers bring fun and more than $48,000 in support

By Colleen Lukoff

Spring and summer were flooded with birthday celebrations, gearing up for completing the Million Dollar Bike Ride and holding other events to benefit the NBIA Disorders Association.

In all, family fundraisers brought in $48,064 for NBIA research, family support services and other programs. Here are the details:

Noelle Kegler of Ann Arbor, Mich., held a fundraiser at the local Pizza House in March, generating $657 for BPAN research. The family has done this before, finding it a great way to raise both funds for and awareness of NBIA.

BPAN RESEARCH DONORS

| Anonymous (2)                        | Brenna Esparza                        | Hank & Deb Nelson               |
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| Jamie Arichuk                        | Gena Everhoy                           | Dominique Zellmer             |
| Eric & Marjorie Backlund             | Jolene Lavegia                         | Megan Ziembowicz             |

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(see fundraising on pg.17)
Dear NBIA family and friends,

Please join me in supporting this year’s Beacon of Hope campaign for the NBIA Disorders Association. I am a monthly giver — aka, a Partner in Hope donor — and am increasing my contribution because I know the value of helping families like mine. I hope you will do the same.

Our family’s journey with an NBIA disorder has been desperately sad and unexpectedly joyful. Our sorrows, like so many of yours, come mainly from the natural progression of the NBIA disorder affecting our daughter. The joy has come from the love, the support and the friendships we’ve found along our way.

Early in January of 2013, my wife, Julie, and I were sitting in a tiny office, crowded with clinicians, reviewing the evidence they’d pieced together. After eight years of dead-ends, our path had led us to the Undiagnosed Diseases program at NIH and to this cramped, sterile office. There, the doctors told us what little they knew. It was terrifying. Josie had mutations on her PLA2G6 gene causing various malfunctions in her nerve cells, which resulted in excess iron in her brain. The condition was exceptionally rare and progressive. They said it would eventually kill her.

Additional details were a blur, partly because of our emotional state, but also because the clinicians had little information to provide. Research was sparse, and we were probably their first exposure to someone with NBIA.

The solitary piece of hope they gave us was a website address, nbiadisorders.org, and news of an NBIA family gathering. They said, “We can’t provide any more answers, but contact the NBIA Disorders Association. They’ve got a conference coming up in a couple months. Attend the conference if you can. They will be able to tell you more.”

We spent hours on that website. We read the disease summary. We read the summaries for other NBIA disorders. We signed up for the conference. We attended the conference. We met the clinicians. We made friends with families who understood our pain and bewilderment because they’d been through it all themselves. We saw the ways that the research was moving forward. And we discovered that potential treatments are making their way into the NBIA community.

It was such a relief to find hope after the diagnosis. The territory where we were headed wasn’t completely uncharted, and it wasn’t just a place of pain. We saw the potential to thrive and be happy.

I have come to feel a certain responsibility for the families diagnosed after us. They will need websites and conferences and research and clinicians. But most especially, they will need hope.

Please help us provide that lifeline. Join us in making a gift to the Beacon of Hope campaign.

Gratefully yours,

Matthew Ritzman
Board of Trustees Member, NBIA Disorders Association

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**BEACON OF HOPE CAMPAIGN**

**for Family Support**

Please become a “Partner in Hope” in our fall campaign as a monthly donor.

Thanks to two generous donors, contributors who increase their monthly gift, along with all new monthly donors, will have their contribution tripled for three months.*

All donations to this family support campaign, which ends December 31, 2019, are appreciated, whether they are one time or ongoing.

You can donate online at [https://nbiadisorders.salsalabs.org/hope](https://nbiadisorders.salsalabs.org/hope) or by returning the contribution envelope provided with this newsletter.

*Total funds available is $9,000
June 8, 2019, was the 6th annual Million Dollar Bike Ride and another successful endeavor by Team NBIA Disorders. Organized by University of Pennsylvania’s Penn Medicine Orphan Disease Center, the event matched money raised by rare disease riders, dollar-for-dollar, up to $30,000. As a result of our fabulous Team NBIA, including riders, volunteers and donors, our organization will soon have $60,561 to spend on advancing BPAN research.

Thanks to Paul Stronski of Arlington, Va., who got a crowd of Stronski men to ride and raise money, including James, Luke, Neil, Patrick, and Paul, along with rider Laura Hodes, also of Arlington. Tommy Ftikas of Cranbury, N.J., Doug Burke of Glenside, Pa., and Steve Florio of Boalsburg, Pa., also rode and raised money.

The riders received support from pit stop volunteers Kristi Florio, also of Boalsburg, Pa., Panayiota Anastasiadis of Neptune, N.J., and Kimberly Burke of Glenside, Pa. Two other volunteers, Christine Tefft and Mara Renz, also of Arlington, worked the NBIA Disorders Association information booth.

This is the third year our team was able to maximize the generous match from UPenn. Details of the call for BPAN research grant proposals are on our website.

Jenny Wildvank of Huntington Beach, Calif., leaped quickly into fundraising for BPAN research upon her daughter Dahlia’s diagnosis on May 1, 2019. She raised $18,772 using Facebook and her Salsa Engage page. In addition, Diana Leano’s sixth-grade class at Joe A. Gonsalves Elementary School in Cerritos, Calif., contributed $850. The students held a bake sale and made an educational presentation after contacting Dr. Susan Hayflick for more information on BPAN.

Amber Denton and Jennifer Sanchez of Houston brought together five Texas families to hold a successful family fun day on June 22 at the local Stats Sports Bar and Grill. This was the BPAN Tribe’s third annual event and included organizers Amber and David Denton; Mary Ann and Jack Ruchirushkul of Cypress; Jennifer and Walter Sanchez; James and Carolyn Bellow Wight of Conroe; and Corbin and Xochitl Galvan Wilson of Nacogdoches.

The group reprised the activities of their first event in 2016, with a King of the Wing competition and a cash prize for the best wing sauce; silent and live auctions, including Astros tickets; T-shirt and bracelet sales; and activities that featured a bounce house, face painting and rock painting. Despite extreme heat and rain, the Tribe raised $10,667 for BPAN research.

Not shown: Tommy Ftikas, Nicholas Decker and Laura Hodes.

Tommy Ftikas and Steve Florio riding for Team NBIA disorders in the Million Dollar Bike Ride.

Five Texas families held a family fun day on June 22 at the Stats Sports Bar and Grill in Houston and raised $10,667 for BPAN research. Their children who have BPAN are left to right: Isabella Wilson, Evalyn Wight, Conner Ruchirushkul, Sydney Denton and Sophia Sanchez.

(see fundraising on pg. 19)
You can honor or donate in memory of a loved one or a friend through a gift to NBIA Disorders Association. The thoughtful people listed below have made a donation on behalf of their friends and loved ones during the last few months.

In Honor Of

BRIANNA AYLESWORTH
Bruce Hesbon & Ellie O’Brien

CHRISTINA BRANT
Vincent Brant

APRIL & TRACIE FLINN
John & Delma Bodwell
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Curtis & Cheryl Ford
Scott & Cindy Heddaeus
Matthew Jenkins
Medtronic Matching Fund Program

BRENDAN WRONKO
James & Sandra Darcy
Meghan Forsatz
Carl & Christine Wronko

In Memory Of

ALYSSA & SABRINA BARBIERO
Norman & Marisa Barnes

AMTUL BHATTI
Rashid & Saliha Bhatti

LEONILDE BRAGA
Anonymous

LOGAN CRAFT
Shirley Santore
Marsha Welsh

WENDY DEVENS
Dennis & Judith Devens

MADISON FREDERICK
David & Mary Ruth

NERISSA GOLDBERG
Candace Kava

BRITTANY LEAP
David & Sharon Baker
Donald & Irene Grass
Deborah Hall
Kelli Bailey Lazarte
Judith Leap
Kelsey Mercuro
James & Anne O’Hara

EALEE DEVOE
Dennis & Judith Devens

DOROTHY WRONKO
Anonymous
Bernie Chrismer

KAIRLE WRONKO
James & Sandra Darcy
John & Mary Ann Daudt
Meghan Forsatz
Robert Sanger & Wendy Solomon
Carl & Christine Wronko

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

Elise Devold
2010 – August 18, 2019

Riley Heasley
October 31, 2008 – July 16, 2019

Samantha Reynolds
February 27, 1991 – April, 2019

Gwen Van Lewe
2006 – July 24, 2019
**MESSAGE FROM THE PRESIDENT**

Our NBIA community received some disheartening news in August. We had high hopes that the clinical trial of a drug to treat one of our most common NBIA disorders would be successful, altering the course of PKAN.

Retrophin’s FORT study for the drug Fosmetpantotenate was designed to see if it improved the lives of those living with Pantothenate Kinase-Associated Neurodegeneration. It was a double-blind, placebo controlled trial — meaning that no one knew who was getting the drug and who was getting a sugar pill. It was not until the study was complete that anyone knew what the findings would show.

Unfortunately, the results showed that the drug had no effect, and there would be no further work on bringing it to market.

I am grateful that Retrophin worked on this and has been a part of our community. The caring employees I have met and worked with these past several years have shown me that they too wanted to help NBIA individuals and cared on a personal level. They have shared with me how sad this news is for them, as well.

Since 1996, when I started our organization, I have been waiting for the day when we could say that there was a treatment focusing on the underlying cause of disease for at least some of our NBIA-affected individuals. But science doesn’t always follow our hopes, and it appears that day is a little further off.

We need to keep the faith and keep plugging away to help make that day happen as soon as possible. There are other projects underway, and science is moving fast, so I hope this is only a bump in the road on the path to a big celebration.

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**Fundraising**

(continued from pg. 17)

Families and friends continue raising funds on Facebook to celebrate birthdays and other occasions. From March through June 2019, we received nearly 500 gifts totaling $28,934. Fundraisers included Gina Engblom, Taylor Kozak, Svetlana Toder, and Jenny Wildvank.


**NBIA Disorders Association is grateful to its supporters for their generosity. We extend our deepest thanks to the contributors listed below who have donated in the past few months.**

Anonymous (5)  Michael Baker International
Amazon Smile  Foundation
Meg Talley Dyer  Jennifer Mickeliunas
Peter Dykhuis  Kimberly Pastore
Angela Faridnia-Brumm  Peter & Jane Sanford
Dennis Gort  Lisa Schafer
Just Giving  Rayburn & Ardell Skoglund
iGive  Andrea Sojak
Kenneth & Lynn Lindebrekke  Rod & Rose Walker

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

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**PARTNER IN HOPE MONTHLY DONORS**

Cyrus & Elizabeth Bradford  Kimberly Mitchell
Douglas & Kimberly Burke  Mark & Cyndy Patton
Warren & Marguerite Cooper  Matthew & Julie Ritzman
Dolores Florio  Mary Ann Roser
Gary & Barb Hampton  Richard & Sandra Savinda
Mark Karakourtis  Roy & Karla Smyth
Colleen Lukoff  Tom & Mary Tapke
Kayinn Mayfield  Richard Tifone
Lynda & Sally Meade  Alex & Angela Wolf

**PATRICIA & KIMBERLY WOOD FUND**

**BEACON OF HOPE CAMPAIGN - Family Support**

Colleen Lukoff  Carmen & Julie Roser  Mary Ann Roser

**PLAN RESEARCH DONORS**

Michael Baker International Foundation  Cooper & Lauren Blackshear

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If you are interested in organizing a fundraiser to benefit the NBIA Disorders Association, contact me at clukoff@NBIAdisorders.org, 760-591-9455 (office) or 760-402-5686 (cell). I am happy to help and can share tools to support your efforts, including our Salsa Engage platform. You can also download our new Fundraising Toolkit from our website at https://www.nbiadisorders.org/images/FundraisingToolkit.pdf.

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**from discovery to cure**
Our Mission:

In our drive to find a cure for NBIA, we provide support to families, educate the public and accelerate research with collaborators from around the world.

NBIA Disorders Association

2082 Monaco Ct.
El Cajon, CA 92019-4235

E-Mail: info@NBIAdisorders.org
phone: (619) 588-2315 fax: (619) 588-4093

Visit our Web site at
www.NBIAdisorders.org

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