



A DAY AT THE FDA

With Mike Kenworthy, Ph.D and Mat Johnson



U.S. Food and Drug Administration
Protecting and Promoting *Your Health*

fellow EPPER, Mat Johnson, joined me. In my 62 years I had never met anyone with EPP outside of my family (I have two cousins with EPP). That was an experience in itself. Those of us with EPP know that we can never explain to someone without EPP what it is like, but listening to Mat Johnson was like reading my own thoughts. Mat and I spoke a couple of times during telephone conferences before we actually met at the FDA in Silver Spring, Maryland. Although our session was scheduled for 10:30 am, we were asked to come early to meet with Dr. Timothy Cote, the head of the Office of Orphan Diseases. I arrived just before 9 am and Mat was close behind. Dr. Cote had just returned from Israel two days prior and was sick the day before, so he was not in good spirits, but after a few minutes listening to Mat and me, he was beaming. He told us that his staff, like all bureaucrats, tends to lose site of their mission and was delighted that we were there to remind them that we are out there. Dr. Bob Desnick, a member of the APF Advisory Board, was also to attend, but his flight from New York delayed him an hour. By the time we were ushered into the conference room over 30 doctors and health professionals were waiting to hear what we had to say. For an hour Mat and I regaled them with the inexplicable world of EPP and the miracle of Afamelanotide during which time we were videotaped. We tried to tell them of the unrelenting pain, the isolation and self-doubt, and ultimate despair. Then we told them about our experience Last Summer and how it had changed our lives. We weren't supposed to directly discuss this, but it just couldn't stay untold – so we did. Dr. Desnick translated what we had to say into meaningful medical speak, but I believe that our words had more impact.



Like most families, we have gone to the beach almost every summer where after 15 minutes, I am banished to the house and sit with my feet in a lobster pot of ice water for the next 5 days “sipping” copious amounts of adult beverages. Needless to say this is hardly endearing to my wife and makes everyone wonder. But last summer, I spent hours on the beach with virtually no pain – a certified miracle if ever there was one. Like most EPPers, I have never told anyone outside of my immediate family about my condition. My colleagues at the office began referring to me as Mr. Tan without ever knowing why. But the most compelling thing that day at FDA was a video that Mat had brought. It was a video of the episode that led to his diagnosis. He was 11 at the time and had gone skiing with his family. The video showed the progress of the effect of EPP over the period of about 10 days. As most of EPPers know, even though the pain has past, we are not a pretty site after 10 days. It had a profound effect on our audience. Drs. Cote and Mueller thanked us for our time and promised that they would advocate our case to the extent they could, but could promise nothing. They explained that there is concern that the drug would be used for “off label” usage and that it would take time and effort to weigh the risks and benefits of a drug such as Afamelanotide. Dr. Mueller told us that she would share the video of our session with the FDA Center for Drug Evaluation and Research. I believe that this was a first and significant step in getting approval for the use of Afamelanotide, but I am not sure that the end is in sight. I live 45 minutes from the FDA campus and with the help of APF will continue to do whatever it takes to gain approval for use of Afamelanotide.

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Mike's Personal EPP experience

My first recollection of EPP occurred when I was four years of age. I was visiting my grandparents in Kansas and experienced a “terrible rash.” No one could figure out what caused it. The best guess was that I had accidentally walked into a hill of red ants, of which there were several, and had been severely bitten. I was treated with a good bath, and some lotion was applied. Within a couple of days, the “rash” subsided. Over the ensuing years, these episodes repeated, particularly in the spring. It was noticed that there was a correlation with the strength of the Sun. So, it was a Sun disease. Somehow my Mother heard of a condition which seemed to be coincidental with mine, Erythropoietic Protoporphyrin (EPP). . . .

READ THE REST OF THE STORY ON THE APF WEBSITE WWW.PORPHYRIAFOUNDATION.COM

READ MATS'S STORY IN THE NEXT NEWSLETTER

Elizabeth Petersen represented the many members who contacted their physicians to arm them with the APF brochures and educational materials. She has been a patient advocate for over twenty years and as such, has helped many porphyria patients. Elizabeth has also volunteered in the APF. Her son, Patrick, is our youngest member at age two.



Editor's note: Elizabeth is my sister. She has been helping the APF since she was a youngster. I can remember when she and my daughter, Lelia, would stuff envelopes, write the addresses and prepare the entire mailing list of the APF. When she got older,

Elizabeth would haul the mailbag to the post office even though the bag was bigger than she was. Now she lives in OK where she and her husband, Chris, have hosted a support group and other education endeavors. Desiree Lyon Howe

Mary Tiege Schellenberger of Lexington, SC has been working overtime promoting awareness within the media, the public, the medical community and



the sports community, namely the Motocross races across South Carolina. Mary's son Miles is a Motocross racer and is quite the rider.

Miles helped his mother promote porphyria awareness at his Motocross event and shared the APF *Porphyria Live* DVD with a reporter with a major TV network. The photo of Miles has NPAW inserted and was passed out at Motocross events across the state. This is our first Motocross. Mary's photo is her with her daughters, her two other treasures.

Diana Ijames has been a very supportive and helpful member of the APF. She also participated in the recent EPP clinical trials with Afamelanotide. Knowing how important it is to enhance awareness of EPP and the porphyrias, she set up a Health Fair at her company,



ACE Manufacturing and Parts Company in Sullivan, MO. Plus, she set up a special EPP booth at the Health Fair where she distributed educational materials, a wonderful idea. Diana and the porphyria booth are shown in the photos at left.

Jana Ebbert is a medical student from Lewisburg, WV. Since she has AIP, she knows the lack of knowledge within the medical community. This is why she passed out the APF education materials to her classmates. When these students graduate, they will have had more porphyria education than most other medical schools thanks to Jana.



Craig Leppert, who is covered up in this photo, was the subject of a recent ABC news story. Craig, a sophomore at Syracuse University, is shown in the photo at his HS graduation as Student Body President. He and his sister, Nicole, have erythropoietic protoporphyria (EPP), which as Craig describes feels like hot wax on your skin, or having your hand cut with a knife and put over a stove when the sun hits. The Leppert family have been long time members of the APF and have been involved in multiple awareness and educational activities, including several television programs, like Dr. Oz and Medical Mysteries. Craig was a participant in the EPP clinical trials. View his story at <http://abcnews.go.com/Health/sunlight-allergy-pouring-hot-wax-skin/story?id=13437546&page=1>

Mary Lipscombe who resides in England joined us in promoting porphyria awareness on her side of the "pond." Mary wrote about our efforts on her website from England: www.barnsleyandfamily.com

Lisa Kancsar serves on the Patient Advisory Committee of the APF and has been a very active porphyria advocate for many years. She has set up education stations at hospitals, facilitated nursing programs and has even put out fund raising bottles to collect money for research. Her daughter has become quite the advocate, too, making presentations at her high school and continued at college.

Nathan Carr encouraged his hospital to place porphyria materials in their library and at Nathan's behest, they not only made the materials available, they sent emails to the medical community reminding them that educational materials on porphyria were available. What a great idea, Nathan!!! Some of you should follow Nathan's example.

Janie Williams was featured in her local news and will continue to see that porphyria is the focus of more articles.

It is not too late too late for you to enhance porphyria awareness in your area. If you would like the APF to send an educational packet to your doctor, you can contact Kate or Yvette at the APF with your doctor's name and address. Remember, too, in June and July you can purchase the *Porphyria Live* DVD and receive one free for your doctor.

NATIONAL PORPHYRIA AWARENESS WEEK (NPAW)



Dr. Joseph Bloomer championed the cause of National Porphyria Awareness Week by participating in six hours of radio interviews about rare diseases but focusing on porphyria. The interviews were aired on nineteen networks around the country and was heard by over 49,000,000 listeners and almost 800 airings. Considering that Dr. Bloomer had just returned from the International Porphyria Meeting in Cardiff, Wales, he had little time to rest before taking up such a monumental task. The interview schedule was placed on the ENEWS but for those of you who missed the interview, Dr. Bloomer's presentation was outstanding!! We hope in a few months to hear or read the text of his interview. He presented the information about porphyria in such a manner that listeners with little medical knowledge could grasp his message about porphyria and rare diseases in general.

Dr. Bloomer, who is a renowned porphyria expert, is Director of the Liver Department at the University of Alabama in Birmingham. He is also a member of the Porphyria Research Consortium, as well as one of the mentors in our Protect the Future program to train future experts. Last year, Dr. Bloomer was awarded the prestigious Presidential Award for Liver Disease, the nation's most coveted award in the field. *THANK YOU, DR BLOOMER.*

Thank You APF Members

Many of our members advanced porphyria awareness through their own projects.

We would like to feature all of your efforts but can only present the following.

Two nine year old girls in different parts of the country were our youngest participants.



has made a great impact on her

Anna Wright is a powerhouse for porphyria awareness. She held a fund raser at her school selling cookies to donate the profits to the American Porphyria Foundation. Then she followed up her fund raising campaign with a presentation to her third grade class about the porphyrias with a focus on EPP. She and her mother, Regina, designed a bookmark noting the APF and distributed it at school and their community. Now her classmates will have an understanding of EPP and what kids endure if they have a photosensitive illness. Anna is one of our APF's youngest members but she has made a great impact on her community, thus encouraging us to do the same.



Alessia Callahan is seen in the photo making a terrific presentation on EPP for her third grade class in Glassboro, NJ. Her school presentation was only part of her effort to advance education and awareness of the porphyrias. She also showed the APF's DVD, *Porphyria Live*, and she distributed porphyria Fact Sheets, EPP brochures and even sun block samples. Like Anna Wright, Alessia is one of our youngest members. Both girls are nine years old and are in the third grade.

It is great news to know that our young members are promoting porphyria awareness and education. Thanks to Alessia and her mother, Jeanine, and dad, Tim, for bringing EPP and the porphyrias to the attention of her community and school. *Thank you, Alessia.*



Joanna Floyd was featured in an outstanding article in her Montgomery County, PA newspaper. Joanna Floyd has a busy lifestyle, like many other 30-year-olds. She is a wife, mother of two and holds down a full-time job as a property manager for a local real estate company. But was diagnosed with Hereditary Coproporphyrria (HCP) when her migraines worsened and she began to have an array of neurological symptoms. Along with the headaches, she developed hot/cold flashes, heart palpitations and stroke-like symptoms. Her speech was slurred and she couldn't walk straight. She was thought to be drunk when she wasn't.

"When I was in the emergency room, I was always asked if the pain could be related to my menstrual cycle. Every time I was asked this question, I told them (the ER staff) this pain was much more severe than menstrual pain," Floyd noted.

Under the care of a family physician, Joanna had every imaginable test and scan, and like other porphyria patients, no diagnosis except a low platelet count, which didn't fit her symptoms. Finally, a coworker suggested a new doctor, who took her ailment very seriously, "You often hear doctors say, 'When you hear hoofbeats, think of horses, not zebras,' but when you have someone come to you who has had a cavalry of horses excluded, you need to think of the zebras." She tested Floyd for three diseases she had not been tested for before — celiac disease, inflammatory bowel disease and porphyria. After all the diagnostic testing, blood, urine and stool, she was finally diagnosed with HCP. Joanna hopes talking about her medical ordeal, the physical and emotional distress she experienced, will spare others the anxiety and frustration she endured for two years. Read the entire story on the web: <http://www.pottsmmerc.com/articles/2011/04/10/life/srv0000011350875.txt>

International Porphyria Conference

According to the US experts, The International Porphyria Meeting in Cardiff, Wales was an extraordinary experience. Porphyria specialists from around the world gathered to share their research and knowledge. Many of them have been colleagues for over thirty years so it is also a time of seeing old friends. The United States experts played a major role in the conference and made a number of extraordinary presentations. Some of our *Protect the Future* (PTF) doctors, were also in attendance. Remember our PTF doctors are in training as our future experts: Dr. Ryan Caballes, Dr. Majed Rizk, Dr. Guilherme Perini, Dr. Tarun Narang, Dr. Ashwani Singal, Dr. Sahid Mittal, and Dr. Bruce Wang.

The venue for the conference was the Readon Smith Lecture Theater of the National Museum of Cardiff. The Scientific Program covered the latest developments in heme related disorders. The meetings included plenary sessions with prestigious speakers and outstanding poster presentations. The attendees also enjoyed contributions from professors in related fields, clinicians, molecular biologists and laboratory sciences. The Scientific Program began after an enjoyable Welcoming Reception at the beautiful Galleries and Grand Hall of the National Museum of Wales. Our APF Scientific Advisory Board member, **Dr. Herbert Bonkovsky**, made the first presentation, the Clinuvel Plenary Lecture, "Acute

porphyrias: Neurologic Manifestations and their pathogenesis.

Other renowned US experts were also presenters:

John Phillips, Ph.D from the University of Utah gave an outstanding lecture, *Intracellular Compartmentalization of Uroporphyrinogen and Uroporphyrin.*

Dr. Charles Parker who is also from the University of Utah, spoke on *Isonicotinylhydrazine (INH) lowers protoporphyrin levels in a mouse model of erythropoietic protoporphyria (EPP).*

Dr. Harry Dailey from the University of Georgia, lectured on *The Bullous Porphyria.*

Dr. Ashwani Singal, who is also a PTF doctor, made the following presentation: *Comparison of phlebotomy and low-dose hydroxychloroquine in the treatment of porphyria cutanea tarda: a prospective randomized study.*

Dr. Charles Lourenço, one of our Brazilian PTF doctors, was unable to give his presentation, *Neurological manifestations in acute porphyrias: review and follow-up of 54 Brazilian patients*, due to work constraints.



1. Bruce Wang and Guilherme Perini 2. Our group of PTF Doctors enjoy the Gala 3. Drs. Ryan Caballes, Tarun Narang and Manish Thapar

Protect the Future

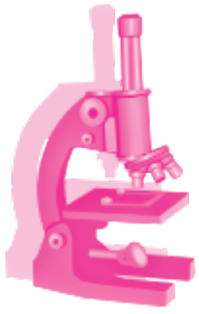
Members of the Scientific Advisory Board of the American Porphyria Foundation are medical pioneers in the field of porphyria and have led porphyria research, testing and treatment for the past thirty years. This prestigious group of porphyria experts have dramatically increased the level of porphyria diagnosis and treatment through their research, physician consultations and publications on porphyria.

The American Porphyria Foundation is acutely concerned about the evolving scarcity of porphyria specialists. Therefore, we created the "Protect The Future" program to attract and train the next generation of porphyria specialists. Our "Protect The Future" program is a major means of expanding the present group of porphyria experts and practitioners while preventing a future lack of experts and researchers to serve the Foundation and its members.

In the past, government funding was available to train young scientists and clinicians in rare diseases. However, this kind of fund-

ing has not been available for many years. We must fund this program ourselves, because our future and that of our children is at stake. Without future experts, who will educate our doctors, perform research, improve diagnosis and treatment or find a cure.

Please make a special donation to the PTF program. At present, the experts are training the following young doctors, Drs, Bruce Wang, Manisha Balwani, Lawrence Lui, Ryan Caballes, Majed Rizk, Brendan McGuire, Sahil Mittal, Ashwani Singal, Gagan Sood, Amanpaul Singh, Guilherme Perini, Charles Lourenço, Bradley Freilich, and Manish Thapar. We are grateful to each of them for their willingness to develop expertise in the field of porphyria. Our future health is in their hands. **We need to support this program to continue their training and add other young doctors. Please make your special gift to the APF and mark it, PTF. Thank you!!!**



Upcoming Research

Upcoming research proposals will be featured in future newsletters, like a [Pilot Study of Circadian Rhythms of Serum Cortisol and Melatonin and Clock Gene Expression in Whole Blood Total Leukocytes of Subjects with Biochemically Active Acute Porphyria](#). The study will recruit clinically stable menopausal women with acute intermittent porphyria (AIP) with elevated urinary delta-aminolevulinic acid (ALA)

and porphobilinogen (PBG). Three healthy controls that closely match subjects' average age, sleep pattern, sleep duration, and caffeine intake will also be recruited. Pre-screened subjects will be requested to undergo frequent blood draws over a 24 hour period. Markers of SCN activity, serum melatonin and cortisol, will be compared between the AIP group and controls. Furthermore, the expression of selected genes in total white blood cells (WBCs) between the two groups will be analyzed.

Modified circadian rhythmicity at the level of either the SCN, or WBCs, or both in AIP subjects would suggest that heme is crucial in the functioning of the molecular clock. A larger study would then be justified to investigate whether medications or drugs that alter circadian rhythmicity also modify symptoms of porphyria. This pilot study attempts to deepen understanding of heme deficiency in relation to AIP and to extend therapeutic arsenal against it.

And

Dr. Herbert Bonkovsky and his team want to conduct research regarding effects of Panhematin use in women who are breast feeding. We feel sure that a number of women have used Panhematin while breast feeding, because of the rather high frequency of acute porphyric attacks in the post-partum period.

And

- Remember the Research Consortium is already at work on their Longitudinal Study.
- To enlist in any research project, go to the APF website and click on Porphyria.
- Pilot Trial of Deferasirox in the Treatment of Porphyria Cutanea Tarda: Deferasirox is a new class of tridentate iron chelators with high affinity and selectivity for iron. The medication is administered orally, which if effective for PCT would make it a more convenient and possibly more tolerable option for patients.

And

Studies in Porphyria I: Characterization of Enzyme Defects, all patients are evaluated for porphyria type and factors contributing to the clinical expression of their particular form of the disease. Testing includes erythrocyte porphobilinogen deaminase, erythrocyte protoporphyrin, plasma porphyrins, and urinary and fecal porphyrins and precursors.

Watch the Enews and the NEWS section of the APF website for more information.

Tell Your Doctors

If you have one of the acute porphyrias, please view this excellent panel discussion and tell your doctors about it.

The Management of Acute Porphyrias: Improving Diagnosis, Treatment, and Standards of Care CME/CE with expert Herbert L. Bonkovsky, MD; and panelists: Owen M. Lander, MD; Maged K. Rizk, MD; and Gale W. Groseclose, RN, BSN.

<http://www.medscape.org/viewarticle/740435>

Need Help???

Many patients who contact the APF have no health insurance or cannot afford their insurance premiums. If this is your situation, you can contact the APF to determine if there is a program that will help you. Contact the APF if one of the following programs may fit your needs:

The HealthWell Foundation® is a 501(c)(3) non-profit organization established in 2003 that is committed to addressing the needs of individuals with insurance who cannot afford their copayments, coinsurance, and premiums for important medical treatments. Our vision is to ensure that no patient goes without health care because they cannot afford it.

As patients are required to pay a larger share of health care costs each year, even many individuals with insurance find health care unaffordable. These patients face challenges affording the treatments they need to fight chronic and life-altering medical conditions. The HealthWell Foundation was established to help alleviate this difficult situation.

The Lundbeck Reimbursement Support and Patient Assistance Program is a free service available to patients, caregivers, medical billing staff, healthcare providers and others who have questions about Panhematin insurance coverage and reimbursement-related issues. The Lundbeck Reimbursement Support and Patient Assistance Program offers help with:

Billing issues — Contact the hotline for assistance obtaining appropriate billing codes required on medical claims or if you need additional documentation to submit with your claims.

Insurance verification — The hotline representative will contact your insurance company to determine in advance how it will pay for Panhematin.

Prior authorization support — The hotline representative can help facilitate the prior authorization process by determining requirements, coordinating paperwork, and following up on the final decision.

Insurer education — Representatives who staff the hotline can help educate insurers about Panhematin to expedite coverage and payment.

Policy monitoring — The program monitors public and private payor coverage policies to ensure you have the most up-to-date coverage information. Representatives will also answer any questions you have about insurance coverage and reimbursement related to Panhematin.

Contact the APF for further information.

An EPP Pioneer, Florence Rollwagen, Ph.D.

My early life was much like others with EPP, painful exposure to the sun, nobody knew what I had, etc. The fact that this was the late 1940s didn't make a difference either, EPP was rarely diagnosed and treated. I was passed from doctor to doctor, various diagnoses were made: She'll grow out of it when she reaches puberty, she's just "acting out", she's missing a layer of her skin. The one I liked best was "if she gets a suntan, the symptoms will go away". I don't have to tell you THAT was a bad idea! So, as the years went by, I learned to cope with my sun "allergy", avoiding the sun, standing in the shade, wearing gloves, I'm sure you know the drill. The worst part, was when I was in grade school, we all had to line up in the schoolyard and march in the class at the beginning of the day and at each recess. Couldn't get out of that, since I didn't have a diagnosis, and schools weren't that "disability friendly" in the 40s and 50s. I spent most of my childhood in some degree of pain. In those days, cameras didn't have flash attachments, so we had to stand in the sun to get a good image. **This photo of me and my little sister says it all.**



I really think that those days gave me the experience of standing up to adversity, and accomplishing my goals in spite of setbacks. There's a sign on the school near my home that reads: "Achievement doesn't let traffic get in the way." Like yours, most of my childhood was spent indoors. We had an old piano in the basement, so I learned to play and discovered I really liked it. I am still playing and having a ball with it!



I went to college to study biology. Girls weren't supposed to be smart, so that was difficult also. The chairman of the biology department was a plant ecologist. One of the "required" courses was plant taxonomy. I spent the whole semester out in the fields and woods identifying plants. What an adventure! Sometimes I wore a ski mask. Sometimes other students would bring back plants for me to identify in the lab. For some reason, I didn't do well in that class – go figure! The rest of college was eventful, I did well and graduated in 1966. The graduation ceremony was outdoors (!). So I couldn't go – another in the long line of events I couldn't attend.

After college, I got a Master's degree in radiation biology (it was the Cold War) and then went to work for a pharmaceutical firm. I worked there for several years, then got a new job at Columbia University where I met my husband-to-be, a resident in pathology. Living and working in New York City was a new experience in freedom for me. The tall buildings effectively gave me a place to walk even during the day! We explored everywhere, walking miles each weekend. I worked several years as a technician in the

Pathology Department at Columbia, then left to enter graduate school at Cornell University (also in New York City). This was in the mid-1970s, and I finished my PhD in immunology in 1979. My husband had to enter the Navy, since he was deferred during the Vietnam war. I took a post-doctoral position at the National Institutes of Health in Bethesda, MD. We had a wonderful time working and exploring the nearby countryside as well as Washington, DC. This city was an entirely new experience. No tall buildings, no public transportation, it was hard for me to get around. Fortunately, my husband didn't like the sun either, so we did woodland hikes, cavern exploration and anything else that kept us covered.

Around 1981, I diagnosed myself by searching the medical literature. The diagnosis was quickly confirmed after a skin biopsy by my dermatologist. I knew there was danger of liver failure, but according to reports, the majority of those occurred in childhood. I was already in my 40s, so I thought I had dodged the bullet. Alas, it was not so. In 1988, I started to feel tired and started to lose weight. Felt increasingly awful late in the year. When I finally saw my doctor, I was diagnosed with liver failure that was pretty far advanced. Since I'm never one to give up, I had worked all through the early symptoms and was in aerobics class two weeks before my transplant. I crashed in late January 1989 and was sent to Pittsburgh by ambulance to wait for an organ. I got a liver in mid-February, just nine days after I was listed. Recovered well and went home in two weeks, and back to work in six. Like I said, I'm never one to give up.

The medications for transplant are daunting and dangerous. The biggest risk is immunosuppression. Without a powerful immune system, one is prone to unusual and dangerous infections. I weathered several, including fungal meningitis, hepatitis, cytomegalovirus, to name a few. Kept going. One thing I learned early on was that if I immersed myself in work, I didn't have time to worry. That strategy has always worked for me, but it's probably not for everyone. It's been 23 years since that transplant, and I'm still going strong. I've retired from my day job, am teaching part-time at two local community colleges. Still having fun, learning every day. Since my retirement, I'm playing the piano better than ever, sleeping better than ever, and my house is finally clean [grin]. It seems as if my whole life was spent overcoming obstacles, career, health, education. I'm definitely a stronger person for it, and although it's been hard, I feel prepared for anything that can come my way. Here's a little poem I wrote:

**Oh, hello again, Death, Back again for another try?
Oh, you'll win, eventually, But not today!**

**RESEARCH IS THE KEY
TO YOUR CURE.**

SUPPORT RESEARCH.

Romanian Reflections

It has been my habit for the last 27 years to always be available for patients no matter where I am and no matter what time it is. The main reason is that porphyria is rare, thus making it hard for people to find help when they are desperate. The office had forwarded a phone call from a woman in Romania, whom I will name Herta. It is not unusual for us to receive calls from all around the world from people with serious porphyria issues. Herta's sister-in-law, we will name Darla, was near death and had just been diagnosed with AIP. Although most of the US experts were in Cardiff at the international meeting, I was able to answer her questions. It seems that her doctors were not informed about porphyria and needed assistance. Aside from basic data that I gave Herta, our APF Office Administrator, Yvette Strange, provided her with the contact info to Orphan Europe, the company in Europe which handles Normosang, the hemin therapy like Panhematin. Herta called me back early the next morning with terrible news. Her sister-in-law was in a coma and had been intubated and her condition was worse. She had already ordered the Normosang, which was to arrive at 3 pm that day. However, she and the doctors had questions regarding the administration of the drug. I promptly contacted Dr. Joseph Bloomer and relayed his instructions immediately. We are anxiously awaiting news of her recovery. Below is the families recount:

Darla was a lovely, loving happy young woman. All this came close to an end a few weeks ago when Darla went to the doctor with abdominal pain, nausea, vomiting and fatigue. Doctors hospitalized her but had no idea what was wrong. Based on the dark purple, red-brown colour of the urine, a young doctor said it must be PORPHYRIA. Darla started to have neurological symptoms, seizures, confusion and hallucinations occurred and the numbness turned into peripheral neuropathy. At the Bucharest hospital, she was tested again. The results shocked the doctors, ACUTE INTERMITTENT PORPHYRIA! It was a pity that this took almost 2 weeks while Darla worsened and was rushed into ICU in a coma.

We were desperate for help, so we called the American Porphyria Foundation and spoke with Desiree. Desiree became our advocate and facilitated our getting Dr. Bloomer's input and Normosang. After Normosang administration, Darla was out of the coma but still in serious condition. The peripheral motor neuropathy progressed quickly to respiratory and bulbar paralysis. We were desperate. We had spent all of our money from the family on the Normosang, the only drug available in Europe. The family struggled to buy it and they managed to buy from Hungary three packs of NORMOSANG and spent all the money the family had or could borrow. Desiree helped us receiving more. **Thank you, American Porphyria Foundation and Orphan Europe.**

The APF is here to help you and others around the world. Please remember that it is your donations that make such life saving events possible and that YOU share in these gratifying experiences.

Congratulations Mira Geffner for being voted onto the board of the National Organization of Rare Disorders NORD. Desiree served on the NORD board for almost 20 years and participated in the adoption of the Orphan Drug Act. Mira has been involved with the APF for many years in a number of capacities. She serves on the Patient Advisory committee and has participated in a number of research projects. Mira is an extraordinary person and is extremely knowledgeable about the porphyrias, so she will be an excellent NORD board member. Fortunately, her presence keeps porphyria in the forefront as she begins her involvement with NORD.

Thank You to APF members, **Amy Chapman, Tracy Yellen and Desiree Lyon Howe**, who were seen and heard on TV and radio in a major promotion for the Porphyria Awareness week. The listeners number in the millions. Perhaps, you heard the message or had a chance to view the Public Service announcements about porphyria. We thank Lundbeck, Inc. for providing this opportunity to broadcast the important messages about porphyria. *Be sure to watch your internet Enews for future broadcasts and other important information.*

In Memory

We are saddened to hear of the passing of our dear friends. Some of their loved ones have chosen to honor a life by making a gift to the APF. We sincerely appreciate their thoughtfulness and desire to help others with the disease. Please join us in thanking: Nancy and Philip Angevine for **Dorothy M. Simko**, Kathleen Toelkes for **Donna Pagano**, Carole and Glenn Kuklewski for **Vince Kuklewski**, Nanette Ragan for **Beki Crowl Huff**, Kaye Isaacs for **Helma Mataxis**, Janice and Richard Hight, Fany Luster, Richard J. Noonan, Janice S. Benicek for **Jane Arbour**, Kathleen and Lawrence Garofalo, Carmen and William Zilch for **Laurie Elizabeth Breiner**, Ellen Johnson for **Hildegard Fermanich**, Joyce Williams for **Deborah Byrd**, Mary Puccia for her son, **Dean Puccia**.

In Honor

Porphyria is an extremely painful illness, and can be life-threatening. The American Porphyria Foundation is working to improve the health of those who suffer with this rare disease by disseminating accurate medical information to patients, educating physicians in appropriate diagnostics and care for the porphyrias, and supporting advanced training for a new generation of porphyria experts. The holidays are a wonderful time to help us advance our mission. Your tax-deductible donation by check or credit card will help us continue our educational work and foster research efforts and the search for a cure. *Thank you.*

Mary Ann M. Kopie for **James, Kathleen, Robert McGuckin**, Judith A. Phelps for **Desiree Lyon**, Allene C. Martin, Eric S. Gray for **Ralph Gray**, Sharon I. Koch for **Matt and Wendy Koch**, Zila and Toma Reichman for **Lielle Judith Ovardia**, Kathryn E. Young for **Linda Manos**, Gerald A. Benedict for **Lisa Kancsar**, Mary Puccia for her sister, **Mary Blanch Hargett**, Desiree Lyon Howe for all of the **Scientific Advisory Board** members and **Protect the Future** doctors who are our hope for the future.



The information contained on the American Porphyria Foundation (APF) Web site or in the APF newsletter is provided for your general information only.

The APF does not give medical advice or engage in the practice of medicine. The APF under no circumstances recommends particular treatments for specific individuals, and in all cases recommends that you consult your physician or local treatment center before pursuing any course of treatment.

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What's New at the APF www.porphyrifoundation.com

Updated Member Stories Section: Full-length versions of the member stories in our newsletter. Please check-out our new **Facebook Groups:** Group 1 – AIP, HCP, and VP; Group 2 – EPP and CEP; Group 3 – PCT.

A free one-credit **Continuing Medical Education** course on the acute porphyrias for physicians is available online. See the website. Also go to www.diagnosingaip.com to learn about diagnosing acute intermittent porphyria.

Is Your Membership Up to Date? Stay current on all the latest news about testing, treatment, and upcoming events. Please take a moment to renew at our website, or call us at the office: 713.266.9617 or 866.APF.3635. *Thank you.*

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