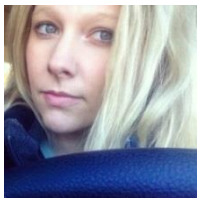

National Porphyria Awareness Week April 11-18, 2015

National Porphyria Awareness Week (NPAW) is the week that the APF collaborates with YOU to enhance porphyria awareness in your communities. You can choose to initiate your own activity to promote porphyria awareness. Some of YOU have participated in health fairs, seminars, fundraising and media events. The APF will supply you with materials to make your awareness activity a success. The Cook family has made such an impact with their Hat Day project that it has become an annual event in their hometown of Vernon, TX. Although her entire story appears in the Members Stories section of the APF website, APF member, Sarah Lee, describes her awareness activities below:

My name is Sarah, and I have Hereditary Coproporphyria (HCP) to be exact. National Porphyria Awareness Week (NPAW) occurs this year April 11-18, 2015. So what is the point in having an "Awareness Week?" It nudges those affected to speak out loud and share their voice. Most of the time talking about our rare disease is awkward, uncomfortable and sometimes a bit scary. NPAW gives us an official reason to engage in a dialog with others in a way we hope won't be judged as "over-sharing" or "attention-seeking." It gives us the opportunity to talk about our cause in a more socially acceptable way. So I am stepping out and sharing a piece of myself in hopes that it might encourage someone else to share their story. The only way we can expect others not to react negatively is by educating them. The person to educate them is YOU--the person they know and care about.

In 1995, my life started to change forever when I began to feel sick. I always felt tired, ill and no longer fared well in the sun. I had pain in my stomach that felt like flaming swords, and I vomited... a lot. I couldn't sleep for days on end and got sudden blinding pain in my head and then pass out. I'd pass out everywhere… in the bathrooms, hallways... heck, I even passed out during a drill on the soccer field.

Then out of nowhere, I felt fine until the illness returned to steal away my happiness, my friends, my energy, my ability to function... my life!! Even after seeing a specialist for two years, I had no answers. I felt sick, alone, and unhappy. Then in 1997, I got so ill at a pool party that I thought I was dying. To no avail, we went to the ER again. However, I was given an appointment with a dermatologist because of my sun sensitivity. I had no idea THIS would be the most significant doctor's appointment of my life. Two weeks later he said, "You have porphyria."

Flash forward to today. I still have all the same sickness, pain, and struggles. The difference is I know the enemy's name. I have educated myself on its tactics, triggers, and I fight back with a special high carb diet, and regular IV therapy. All of this is better, but not enough for me. I still hope for a cure, if not for myself, then for my family, because it is hereditary. But finding cures requires research and that requires funding. Unfortunately, getting that for a rare disease isn't easy. So we have OUR own Foundation called the American Porphyria Foundation (APF). The mission is to educate patients and physicians, to promote porphyria awareness and to support research. Together, we the APF, help with own fundraising, educating, talking to politicians, watching the drug laws, and supporting each other and others with rare diseases. That is why I also participate with the Global Genes Project that unites people with Rare Diseases and become a voice of many instead of a voice of a few. HOPE - It's in my Genes! NPAW is coming up soon, and I plan to advance awareness. It is my responsibility, no one else's, to educate the masses and turn that negativity into understanding. I Care About Rare! Do you?

No pressure, but if you would like to make a donation to the American Porphyria Foundation to help with life changing and even life-saving research, please contact www.porphyriafoundation.org or call 800.APF.3635. Thank you.

FINNIAN PROMOTES PORPHYRIA AWARENESS

Finnian, nicknamed Finn, can show us a thing or two about promoting porphyria awareness. He wears his American Porphyria Foundation bandana proudly. The APF gave bandanas to all the pets who entered the APF Pet Beauty Contest a few years ago. Finn is a 7 year old yellow lab. His mom is APF member, Kelsey Castro who says of Finn,

"He is my best buddy! He is always by my side, especially when I'm not feeling well… I think he can sense it. I got him when I was 14 and in high school, and I'm now in college. Finn has been my best friend through lots of hard times. His favorite thing to do is go on hikes at a local nature reserve by my house and curling up by our wood stove in the winter."

Editor's note: We also ask you to take every opportunity to spread the word about porphyria. Your effort can be as simple as purchasing an APF T-shirt or giving out medical information to your doctors or showing the APF Porphyria Live video to your family and friends. Every community needs to hear about porphyria, and you are the person to do that in your community. Please contact the APF for supplies.

The APF has been asked to host another PET BEAUTY CONTEST. Watch for an update in the summer newsletter!

THE SHADOW RIDE Who doesn't love a cowboy love story? You will especially love this one, because it involves APF member, Scott MacMeeken, and his beautiful bride, Michelle. After they married, Scott saw firsthand how difficult it was for Michelle to live with Erythropoietic Protoporphyria (EPP), a severely photosensitive porphyria that causes pain and swelling. Scott desperately wanted to help Michelle, but soon discovered that he couldn't find a doctor who had even heard of porphyria, much less treat the disease. After learning that people with all the porphyrias suffered with different but painful symptoms, Scott decided to help the American Porphyria Foundation advance porphyria education and awareness. Together they could bring porphyria out of the shadows and into the light.

Scott initiated **The Shadow Ride,** a horseback ride to spread awareness along the American Discovery Trail from the Delaware coast to northern California. As a forerunner to the 2016 ride, we will host a Kick Off ride for the Shadow Ride and National Porphyria Awareness Week, Saturday, April 11, 2015 on the famous Drummond and Hull ranches near Tulsa, Oklahoma in Pawhuska. There will be fun for everyone: **live music, games, miniature donkeys to ride for the kids, the opportunity to meet other porphyria friends and Mary Hull's delicious Bar-B-QUE**…plus, of course, **The Shadow Ride.** Save the date, put on your cowboy hats, boots and jeans and join the fun. You can hang out all day if you like from 10 AM till 7 PM or come for the afternoon Bar-B-Que and activities starting at 2 pm. See: www.theshadowride.com or www.porphyriafoundation.com Call the APF at 1.866.APF.3635 for a map to reach the Hull ranch near Pawhuska, OK.

PATIENT EDUCATION MEETING Patient education meetings are educational and fun! Note the day following the Shadow Ride on April 12, 2015, the APF will also host a patient education meeting in Oklahoma City, OK with porphyria expert, Dr. Sylvia Bottomley. The meeting will take place from 2-5 pm at the fantastic National Cowboy and Western Heritage Museum. After the meeting, Dr. Bottomely's husband will lead a tour of the museum for those who are interested. Patients, friends and family are welcome to join us for a presentation and question/answer session. Our members enjoy these patient meetings because they are not only a time for learning about the porphyrias, but they also provide us with an opportunity to make new porphyria friends. Meeting other people with your disease and sharing your stories, coping tips, etc, a very positive experience. Our motto at the APF is: " Those who know the most, do the best!!! The museum address is 1700 Northeast 63rd St, Oklahoma City, OK 73111. Please let the APF know you are coming. You can email porphyrus@porphyriafoundation.org or call us at 866.APF.3635. If you forget to sign up, please come anyway. Y'all come!

DR SYLVIA BOTTOMLEY will be the featured expert at the Oklahoma City Patient Education Meeting. She is a renowned porphyria expert and hematologist in Oklahoma City, Oklahoma. Dr. Bottomley received her medical degree from the University Of Oklahoma College Of Medicine and has been in practice for 56 years. At present, she is with the Medical Service, Veterans Administration Hospital, OKC. Early in her career, she received a special Leave of Absence to study porphyrin biochemistry and methodology at University of Minnesota, laboratory of C.J. Watson, M.D., who was an early pioneer in the porphyrias. From that period to date, Dr. Bottomley was instrumental in the early studies on hemin therapy and erythropoietic protoporphyria.

She can be seen on the APF *Porphyria Live* DVD and she assisted the APF in developing educational materials and consulting with primary care physicians. She was selected to write the chapter on the porphyrias in the most prestigious medical textbook on hematology, *Wintrobs Clinical Hematology*. She also authored the section on CEP and EPP in the Porphyrin Handbook and she wrote a journal article on Acute Intermittent Porphyria in Genetics in Medicine, the Official Journal of Medical Genetics and Genomics. As an emeritus professor, Dr. Bottomley continues to teach hematology-oncology fellows and advises physicians on patient problems regarding porphyrias and sideroblastic anemias. She remains on staff at the VA Hospital, reviews manuscripts for journals and maintains her research lab, pursuing small projects and collaborating with colleagues elsewhere in searching out the genetic causes of sideroblastic anemias.

The Bottomleys are a close-knit family. Her daughter is a radiation oncologist in OKC and her son is an entrepreneur for software companies. Plus, she has three grandsons. As time allows, Dr. Bottomley enjoys classical music, reading, snow skiing and traveling in conjunction with international conferences.

Dr. Bottomley will be the featured expert at the Patient Education Meeting in Oklahoma City, OK on April 12, 2015 from 2-4 pm following the Shadow Ride on the 11th of April. The meeting will be held in the famous Cowboy Museum 1700 Northeast 63rd St, Oklahoma City, OK 73111. Her husband, who gives tours of the museum, will be available to give the attendees a tour of the museum.

APF PRESIDENTAL AWARD The APF awards the Presidential Award each year to people who have assisted the APF in achieving the mission to fund research and provide education, awareness, and support to patients, their families and their doctors. This year's recipients are Amanda Boston and Mike Kenworthy for their extraordinary efforts in research, awareness and education. We sincerely appreciate their great work on behalf of all people with porphyria.

CONGRATULATIONS AMANDA BOSTON Amanda is a one woman dynamo when it comes to promoting porphyria awareness and helping other patients. In the left photo, Amanda is delivering APF brochures and medical information to her local hospital. We ask each of you to help promote awareness in your local community, including educating your own doctors. The reward from your effort is very great and it may help your own children one day. To recap Amanda's story, in 2011, she began having severe abdominal pain and terrible skin lesions. No doctor was able to diagnose her even after she experienced bad seizures for which she was given an unsafe drug that sent her into a severe attack. Shortly thereafter, she had another attack that was so dreadful she couldn't walk, had blisters that wouldn't heal and was put in the hospital, paralyzed for three months until a resident doctor thought of porphyria. Even after her diagnosis, she spent eight months in the hospital progressing from paralysis to a wheelchair and then to a walker. Amanda is quite a survivor. Now she is out spreading information about porphyria and has even participated in three research projects. She receives Panhematin® every other week to control the porphyria but still has attacks at times. She also is a Facebook Administrator on the five APF groups. Amanda says," I want to help wherever I am needed. "**I may not be where I want to be, but I thank God I'm not where I used to be."**

CONGRATULATIONS MIKE KENWORTHY Mike Kenworthy, PhD is also receiving the APF Presidential Award. He has been an outstanding advocate for porphyria, specifically in the area of EPP education and research. Mike is a rocket scientist, so that he not only brings his experience with EPP, but also his scientific understanding of the disease. We have often asked Mike to write letters of explanation for key people in approval process could better understand the disease from a patient perspective. He also provided the APF with exceptional and effective letters for the FDA, Congressmen, EMA (European Medicine Association), and other regulatory offices. His letters were so effective that the specific offices made note of them. Many of you have read about his many efforts to assist us in gaining EMA approval of Afamelanotide/Scenesse, the new treatment for EPP and his efforts to gain EMA and FDA approval. Mike joined other EPP people in three visits to the FDA and participated in the film the FDA posted on EPP. Mike also participated as a patient volunteer in the clinical trials. Furthermore, he was very instrumental in securing Phase III clinical trials when the FDA was putting them off a year. As long as it has taken to get approval, it would have been even longer had it not been for Mike. Aside from those feats, Mike asked his swim team to donate their charitable donations to the APF. Congratulations Mike!!! And Thank YOU!!!

ALNYLAM RESEARCH ONGOING Alnylam Pharmaceuticals new RNAi therapy for acute porphyrias is already in the initial natural history studies. Our research motto is "Remember, research is the key to your cure." You are needed to participate in these studies. Joining a research project is simple. All that is required is that you have a confirmed diagnosis and then contact the APF. We will put you with a research team. The research team will go over details with you. If your case fits the parameters, you will be flown to the closest Porphyria Center where you will have an examination, donate your blood and answer detailed questions about your attacks of porphyria. Your flights and expenses related to the research study are part of the grant and are not charged to you.

It is not difficult to do, but it is very important for the research and future of the new therapy they have developed. Please volunteer and be a Medical Hero.

SAVE THE DATE: International Porphyria Patient Day and Porphyria Conference

We are happy to announce that the next International Porphyria Patient Day will be held in conjunction with the International Porphyrins and Porphyrias Conference in Düsseldorf, Germany. The conference will take place from September 14th to September 16th, and the Patient Day will be held on Sunday, September 13th, 2015. Presentations on each of the porphyria will be featured, including the new emerging therapies. The International Conference is a gathering of porphyria experts and researchers from around the world. This conference provides them with the opportunity to share their research and discuss the present and future therapies, case studies, etc. This is an exciting meeting that is held every two years. **SAVE THE DATE.**

PATIENT EDUCATION MEETING Hosting patient education meetings are one of the most important functions of the APF. The attendees hear presentations from renowned porphyria experts and even have the opportunity to ask questions about their own cases. The meeting provides the rare event of meeting other people with porphyria, which for most attendees is a very enjoyable experience. We encourage you all to attend the meeting closest to you. The next one will be in Oklahoma City, OK.

The APF held six patient meetings in 2014 and hopes to host even more this year. Watch our newsletter, mail ENEWS carefully for the time and location. If you do not receive the APF ENEWS, please contact the APF and give us your email address to be added and receive the ENEWS promptly. If you would like to host a patient education meeting in your area, please contact Jessica at the APF.

PORPHYRIA TRAINING WEEK AND THREE NEW SATELLITE CLINICS Forty experts, PTF trainees and research coordinators met in Galveston, TX at the University of Texas Medical Branch Porphyria Center for a Continuing Medical Education (CME) Course and training seminar on porphyria. The three day meeting was packed with intense porphyria education on each type of porphyria, as well as time with patients to learn from them. The CME course was also the first training sessions for our newest *Protect the Future* (PTF) doctors who participate in the PTF training program to train the next generation of experts. The agenda included intensive presentations on each type of porphyria, heme biochemistry, treatment for each of the porphyrias, upcoming treatments, the pathology of porphyrias and other related subjects. Important to the meeting were the patient presentations. Patients with different types of porphyria talked to the medical professionals about their personal experience with their type of porphyria and answered a host of questions from the attendees. The attendees were also awarded Continued Medical Education (CME) credit for their participation in the course. It was an exciting and hopeful experience to know that our health is in the hands of these brilliant young doctors and nurses. Thank you all for your participation.

Also three new satellite porphyria clinics have been created. Diagnosis and treatment of all porphyrias will be available at these new clinics. They also will be involved in the research with the Porphyria Consortium. This is exciting news for us all. If you want to make an appointment with one of these new doctors, please contact the APF for the contact information.

- The Miami Clinic is headed by hematologist, Dr. Cynthia Levy and Coordinator Odalys Rodriguez Bravo;
- The Seattle Clinic is headed by Dr. Sioban Keel and Coordinator Regina Delrosario;
- The Cleveland Clinic center is headed by Dr. Angelika Erwin and Coordinator Allison Schreiber.

YOU ARE IMPORTANT Every research volunteer is a "Medical Hero." Tara Cantley is one of the most recent patient volunteers to enroll in the Longitudinal Study, Panhematin Study and the Alnylam Natural History Study of Acute Liver (Hepatic) Porphyria. She felt that by volunteering, she would make a huge difference in the lives of patients now and in the future. Tara flew to the University of Texas Medical Branch in Galveston, TX to meet with one of the leading experts on porphyria, Dr. Karl Anderson and his research team. After Tara *(left in photo)* completed her research requirement, Jessica, APF Director of Patient Services, visited Tara in the hospital. Jessica *(right in photo)* enjoyed hearing about Tara's participation as a research volunteer and her efforts to promote awareness and porphyria research. Jessica, who is new to the APF office, also had the unique opportunity to meet Dr. Anderson and the nurses for the first time. Tara encourages others to volunteer and wants everyone to know: "Research is the key to our cure!" Thanks to all of the research volunteers.

WHAT A COINCIDENCE Pierre Mouledoux had an very unusual experience recently. As you may remember, Pierre was responsible for changing the legislation in Louisiana to assure people with EPP could have automobile window film to protect them from the sun. When he was stopped at a road block, a Louisiana State Trooper saw that he had window film on his car. The trooper told Pierre that his medical exemption didn't cover the windshield. In turn, Pierre promptly gave him gave him the story of the law change and mentioned the unfortunate "vampire publicity" that followed. The trooper was very surprised, because just that morning at the pre-shift change meeting, he and his colleagues had discussed EPP, Pierre and the legislation that provided auto film for EPP people. The trooper couldn't believe that he was the one who stopped Pierre!!! If you have EPP or PCT or other photosensitive porphyrias, you may be interested in gaining permission to use this auto window film in your state.

DR BONKOVSKY CREATES HEMATIN Dr. Herbert L. Bonkovsky, Professor of Medicine and Molecular Medicine and Translational Science, Wake Forest University School of Medicine, Winston-Salem, developed the first hematin in the world. The article below is a summary of his great discovery. You can read the entire fascinating article on the APF website. Recently, I asked Dr. Bonkovsky to write his personal reflection of his creation of hematin therapy for acute porphyrias. Since I was one of the first patients treated with hematin during a life threatening attack and have seen thousands of patients saved by this treatment, I knew our readers would be interested in his account.

Dr. Bonkovsky moved to the National Institutes of Health, Bethesda, MD, as Clinical Associate in the Metabolism Branch of NCI (1969-1971), where he worked in the laboratory of Donald P. Tschudy, MD, one of the 20th century pioneers in the study of clinical and experimental acute porphyrias. Dr. Tschudy had shown a few years before that there is marked up-regulation of hepatic ALA synthase in subjects with biochemically active AIP and that this abnormality is central to the overproduction and over excretion of ALA and PBG, which are key biochemical hallmarks of AIP. (It was only a few years later that Dr. Bonkovsky and others learned that the primary metabolic defect in AIP was decreased activity of porphobilinogen [PBG] deaminase, the third enzyme of the heme biosynthetic pathway.)

He says, *"Based upon studies, as well as those of others in the Tschudy lab and elsewhere, in several experimental models of AIP, we had developed the concept that there is a small but critical regulatory heme pool in hepatocytes and that this pool exerted a key regulatory role on ALA synthase, the first and normally rate-controlling enzyme in the heme synthetic pathway. We and others had recently shown that the administration of heme to cell cultures or experimental animals with chemically-induced porphyrias, in which there was an uncontrolled induction of ALA synthase, was able quickly and profoundly to end this induction and its attendant overproduction of ALA and PBG. We since have uncovered at least four ways by which heme decreases the levels and activities of hepatic ALA synthase [also known as ALA synthase-1], namely, by decreasing transcription of the gene, by decreasing the stability of the mRNA transcripts of the gene, by blocking the uptake of the enzyme protein into mitochondria, and by decreasing the stability of the enzyme within the mitochondria. The bottom line is that, under conditions of relative heme deficiency in liver cells, there is up-regulation of ALA synthase-1, whereas, under conditions of heme excess, there is down-regulation or repression of ALA synthase-1.*

While we were working on these experimental and basic research studies, we also were caring for patients with acute porphyrias in the clinical center of NIH on campus in Bethesda. One unfortunate young woman was suffering from a very severe attack of AIP. She had been in the hospital for some time, and despite our best efforts to help her, she had become totally paralyzed, was dependent upon a respirator (because of profound weakness to the point that she was unable to draw breath), was severely hypertensive, and had developed acute on chronic renal failure. In short, this woman was dying of an unrelenting AIP crisis. Because all other treatments had been exhausted, I proposed to Dr. Tschudy and other senior leaders of our Branch that we should try giving her heme intravenously, in hopes that some of it would find its way into her liver cells and perhaps other cells that were deficient in heme and were over-producing ALA and PBG, one or both of which were felt to be the most likely neurotoxins in AIP and the other acute porphyrias. We had nothing else to offer our patient, and, after careful consideration and discussion, and with the informed consent of the woman and her family, it was agreed that I would purify and crystallize heme from outdated human red blood cells, and that we would infuse this heme into our patient.

Accordingly, I obtained several pints of outdated blood from the blood bank of the NIH clinical center, and I set out to purify and crystallize hemin chloride from these units of blood. The purification involved several steps, including eventual crystallization of the product from boiling acetic acid. I worked all day and all night, and by the next morning, I had beautiful, extensively washed and purified crystals of the desired product. The final yield was about 1,000 mg of hemin chloride. Of course, we had no real idea of what the right dose would be for patients with AIP, but, based upon the results in rats, we believed that 3-5 mg per kilogram body weight would be reasonable. Our patient weighed about 60 kg, so we decided to administer 250 mg every 12 hours in our initial study. The next challenge was to redissolve these crystals in an aqueous solution, suitable for administration to humans. Hemin is not very soluble at physiologic pH [7.4], but it is soluble at high pH. I decided to dissolve the precious crystals into a solution of 1% sodium carbonate, which has a pH of about 10-11. I was able thus to dissolve the 250 mg [about 10 ounces] of hemin in 25 mL of the sodium carbonate. Heme at this pH is actually in the form of hydroxy-heme, with OH^- anions replacing the Cl^- ions, and this form of heme had been named hematin by Hans Fischer, MD, PhD, the outstanding pioneering German chemist, who had won the Nobel Prize for chemistry in 1930 for his work on pyrroles, porphyrins, and the chemical synthesis of heme. Thus was hematin for human use first born. After dissolution, the pH was adjusted downward slowly and carefully with hydrochloric acid to a final pH of 7.5, and the final solutions were put into brown vials for immediate administration. We felt the latter was important because it was known that hematin in aqueous solutions undergoes rapid 'aging' reactions, with formation of polymers and degradation products that may not have the desired therapeutic effect and that also may cause adverse effects. The bio-

chemical effects of the first four infusions, 250 mg given every twelve hours, on April 13 and 15, 1971, were dramatic, with marked decreases in urinary and serum ALA and PBG. Unfortunately, in this woman who had been severely ill for months and, by now, was totally paralyzed, we did not observe clinical improvement.

Nevertheless, it seemed worthwhile to treat her with additional doses of hematin. Thus, for the next several weeks, I spent all day and most of the night, purifying, crystallizing, and preparing hemin chloride for further infusions. We gave several more infusions of hematin to the patient during the next month or so, with further strongly positive effects of decreasing levels of ALA and PBG, both in the serum and in the cerebrospinal fluid. [By this time, due to the ravages of AIP with severe systemic arterial hypertension, acute on chronic renal disease, and superimposed sepsis, the patient had developed severe kidney failure and had stopped making urine. We reported these findings, so that other physicians and scientists around the world could benefit from our experience and could further test the effects of hematin earlier in the course of acute porphyric attacks.

I finished my two-year appointment at NIH in June, 1971, and went to Dartmouth Medical School where I continued training in Internal Medicine and Gastroenterology and joined the full-time faculty in 1974 as founding chief of the GI service at the affiliated VA Medical Center at White River Jct, VT. I spent 1973-1974 training in the young subspecialty of Hepatology, at the Yale Liver Unit, working especially with Gerald Klatskin, MD, and Joseph Bloomer, MD, where I was fortunate to make another seminal contribution to the study of porphyrias, namely, demonstration of decreased activity of the enzyme ferrochelatase, the final enzyme of the heme biosynthetic pathway, as the principal defect in another, cutaneous form of porphyria called erythropoietic protoporphyria. But this is a topic for another reflection.

My successors at the NIH (Drs. Magnusson, Lamon, and others) and physician-scientists elsewhere, especially in Minneapolis at the Watson laboratory [Drs. Pierach and Watson], New York (Drs. Anderson, Kappas, and Sassa) and San Francisco [Drs. Bissell and Schmid], came to use hematin early in attacks of AIP with excellent results. Eventually, in the early 1980's, thanks in no small measure to the drive and determination of Desiree Lyon, Executive Director of the American Porphyria Foundation, who required prolonged admission to the NIH Clinical Center and prolonged treatments with hematin for severe recurrent, nearly continuous attacks of AIP, we convinced Abbott Laboratories of Chicago, a leading producer of blood products, to produce Panhematin®, a stable lyophilized [freeze-dried] form of hematin dissolved in sodium carbonate. It was the first of many orphan drugs approved for use in rare diseases by the US FDA. Now, 44 years later, Panhematin® (or its first cousin heme arginate (Normosang), available in Europe, South Africa, South America, Australia) remain as the clear treatment of choice for attacks of any of the four forms of acute porphyria. It has also had beneficial effects in other conditions of heme deficiency, such as lead poisoning, hereditary tyrosinemia type 1, protoporphyria, and congenital erythropoietic porphyria, but its chief role continues to be the treatment of acute attacks of porphyria, due to AIP, hereditary coproporphyria, variegate porphyria, and porphyria due to severe deficiency of ALA dehydratase.

It goes without saying that I am both fortunate and humbled to have been present at the creation of heme as a treatment for human disease and to have helped now thousands of patients to be relieved of much pain and suffering. The severity and prognosis of acute porphyric attacks are now considerably better than they were prior to our development and use of heme for acute porphyric attacks. Our patients have reported that hematin is the best and most effective available treatment for acute porphyric attacks.

EVELYN JACOBUCCI AIP Talks about Pain.

Tonight as I sit watching Thomas the Train with my babies rocking them to sleep, there is an internal battle waging, and I am it's unwilling participant. I do not say "victim," because I will never let it win! Tonight under the sweet, peaceful, sleeping baby, under the rumpled shirt and tough skin, there is a fire-breathing dragon clawing his way out of my gut. The burning pain sears every nerve. It is enough to bring me to my knees. It's claws rip at my internal organs until it sweeps my breath away. I try to stand but the agony has worn me down. Somehow a cement mixer has poured it's heavy weighted load into my veins. I decide the floor is a safer option for now and lay down. Suddenly the wet cement has moved to my chest and now the dragon is attacking there as well. Still, my babies are watching. "Deep breaths, Ev, you can do this. You deal with this at least twice a month and sometimes more. It just came on so suddenly! I didn't have time to prepare!" I'm arguing with myself... oh dear now I think I might be crazy. I try to turn over to smile at my little girl but the knots that are tied in my arms and legs are making every movement, every breath excruciating. I think I scared my angel because now she is crying. Through the agony, I rise to my feet and pick her up settling us both in the large rocker. Dear God, when will this end. Another IV another round of meds, another frustrating hospital visit where the doctors treat me like a child or a drug addict, another few hours lost with my family in a hospital bed. If only I had something visible to show them how all-consuming this pain is, how my life grinds to a halt, how my children live in the shadow of this dragon who is constantly waiting at my doorstep.

PORPHYRIA RESEARCH CONSORTIUM The PRC met in conjunction with the training week. We announced in the last newsletter that the Consortium had won one of the few National Institutes of Health (NIH) Rare Disease Research grants. We are very grateful to Drs M. Bissell, J. Bloomer, J. Phillips, C. Parker, H. Bonkovsky, K. Anderson and R. Desnick for saving our lives. I hope you will show your thanks by volunteering for a research project. Their research involves a number of projects, so you can choose what you would like to do. If you have a definite diagnosis of any of the porphyrias, you are needed. The volunteer process is easy. Contact the APF and Natalia or Jessica will place you with the appropriate research team. Make a difference. Be a Medical Hero!!

1. The family study requires that one person have a definite porphyria diagnosis. If you have relatives who have not been tested, the family can join the study. It only requires that they donate their blood and answer a number of very pertinent questions. Each participating family member will also have a DNA test. If the participants have insurance, the insurance will be billed. There will be no extra out of pocket costs to the research volunteers. If they do not have insurance, then the test will be free. Volunteering is very simple. This research is being conducted at San Francisco Porphyria Center with Dr. Montgomery Bissell.

2. There is also a Longitudinal Study. Again, having a confirmed diagnosis is essential. In this study, you are required to answer a lengthy list of questions twice a year for five years. The research team will track your illness and make important determinations from these well designed set of questions.

3. The Alnylam study has begun. This is a natural history study for their new RNAi treatment. You will be flown to one of the Porphyria Centers closest to you at no cost to you. The research team will draw your blood, perform an examination and ask pertinent questions. This part of the research only takes one day of your time.

4. Although Panhematin® has been a very successful treatment for many years, it is now being used extensively to prevent attacks. A study will be useful for physicians who want more information on this and other questions. The study is being conducted at the University of Texas Medical Branch in Galveston, TX. You will be flown to the center at no expense to you and will participate in the study for a number of days.

5. There is a PCT specific study being conducted at the University of Utah.

IN MEMORY We are saddened to hear of the passing of dear family members and friends and offer our sincerest condolences. Please join us in thanking these friends and family for their donations to the APF in memory of their loved ones: *Dr. Susan Engel for **Lee Engel**; Grace Ann Feczko for **Robert Zieles;** Mary Alfrey for **Mary Jane Alfrey;** Michelle Letkemann for **Walter F Cernik;** Circuit Riders Class FUMC, Ruth Draper, Debra Reese, Louis and Janie Miller, Jacob, Jean and Finn Pond, for **James Bradley Crelia;** Catherine Gear for **Paul F Sheehan;** Valine Jensen, Linda Peterson, and Kathleen Toelkes for **Donna Pagano;** Edwin Brandt for **Delores Brandt;** Desiree Lyon Howe for **Merri and Sarah Davis;** Norma and Ronald Brown for **Matthew Brown;** Diane Levere for **Dr. Richard D Levere;** Rosemary M Houlihan for **Vera Ferris;** Ralph M Gray for **Fred L Gray;** Kyle J Helvig, Rod Jones, William Grant for **Grace Warfield;** William H Key for **Velma Key;** Donald Johnson for **Peggy Lewis Johnson;** Vickie and Matthew Gehm for **Judy Coley;** Pamela Duren, Dena and Family, Dianne and Karl Wolter, Jamie and Thomas Williams, Julia, Gary, Christian and Lauren, Hutnik, Grace and David Smith, Dawn and William Sherman, Sandra L Parks, Tyler Parks and Troy Parks, Susan Jones, Rebecca Perkins, Elaine J. Mickelson, Jill and David Mittleman for **Lisa Marie Grewal.***

IN HONOR We also thank those friends and family who made a donation to the APF in honor of their loved ones. Since the APF is an important part of their lives, we know that they are most appreciative as are we at the APF: *Elaine Smuczynski for the **Smuczynski Family;** Margie and Gary Lewis, Holly Weinstein and Howard Klein for **Jonathan Turell;** Anne Johnson for **Candace M Johnson;** Mark and Nancy Blum and Jennifer R Ewing for **Desiree Lyon Howe;** David Russell for **Craig and Nicole Leppert;** Cheryl Harriman for **Jeffrey Pradovic;** Nicholas Guanciale for **Kathleen Guanciale;** Gene Ackerman for **Troy Ackerman;** Carol and Claude Gaudette for **Tristen Gaudette;** Steve Andrade for **Josephine Dzygala;** Susan L Cerkoney for **Fred, Jason and Stephanie Cerkoney;** Linda Nagin for **Melissa Nagin;** Michelle Letkemann for **Frank Cernik;** Kathleen E Venter for **Patricia Wright;** Michael P Farina, Margaret E Whittenburg for **Jocelyn and Jamie Whittenburg;** Connie and Martin Helleson for **Jennifer Blackburn Streeter;** Suzanne Michael Godsted for **Tracy Yelen;** Rona Farley for **Dale Hawk;** Daniel Diddlemeyer for **Florence Diddlemeyer;** Rod Jones for **Leann Warfield;** Kathleen A Shiel, Allen C Martin, Myrna C Cartledge, William A Gray, Sara E Collier, Paula M Hendrix, Lori Hansen, Robert DuBord, Mary Frances Donnelly, Ian Gray, Larry and Sarah Pritchard, Jolynn and Keith Foldesi, Ruth Wilson and Robert Hendrix for **Ralph Gray.***

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