



Research: The Key To Your Cure! The *Porphyria Research Consortium* has won funding from the government to perform research, which it is on-going NOW. However, the researchers (shown below) need far more volunteers to continue their important research. If not, the government will not renew the funding, thus, proving again that the patient is the most important key in this process. This is your invitation to make their research a success through your participation as a patient volunteer!!!! The following information contains some of the research and instructions for your involvement.

First, everyone who wants to participate in research, should join the NATIONAL PORPHYRIA RESEARCH REGISTRY by going to the APF website and clicking on "Porphyria Registry" and following the directions or see: <http://rare diseasesnetwork.epi.usf.edu/porphyrias/register/index.htm>. A participant has the right to non-disclosure of their confidential, genetic information. The registry is anonymous of charge. After you join and answer the questions, a member of the *Porphyria Research Consortium* from one of the six Research Centers will contact you personally and answer your questions. You also can contact the APF for questions.



L to R: Dr. Herbert Bonkovsky, Dr. Karl Anderson, Dr. John Phillips, Dr. Robert Desnick, Dr. Joseph Bloomer, and Dr. Montgomery Bissell (below) are members of the *Porphyria Research Consortium* performing a major **Longitudinal Study**. They are in great need of research volunteers. You will not be taking an experimental treatment in these studies, rather, you will be asked questions and your bloods, etc. will be studied. The first step with all the research projects is to join the *Porphyria Research Registry* to better enable the research team to contact you and keep you updated. The process is easy and not time consuming, however, if you would like to have some help, just contact the APF office, 1.866.APF.3635 or email: porphyrus@aol.com.



Dr. Montgomery Bissell, Chief of Gastroenterology at UCSF Medical Center and Director of the National Institutes of Health (NIH) - supported Liver Center, is a consortium of 40 independently funded researchers who study the liver and its diseases. Dr. Bissell is also a renowned porphyria expert and member of the NIH supported *Porphyria Research Consortium*. His study will establish 'who are the porphyria carriers in the family' and 'who are not.' Carriers will be offered enrollment in the **Longitudinal Study**.

1. They need to know the person(s) in the family who have confirmed porphyria including genetic diagnosis (the "index" cases). Hopefully those people are already enrolled in our **Longitudinal Study**, which means that we have the documentation. If not, they will need copies of test reports. Once the diagnosis is confirmed, enrollment in the **Longitudinal Study** will be offered along with the **Bissell Research Project**.

2. A study participant can be any first degree relative of an "index" case (sibling, parent, or child). She/he must be age 15 and up, must be able to consent (or have a parental consent) and must not yet have had genetic screening for the porphyria gene in the family.

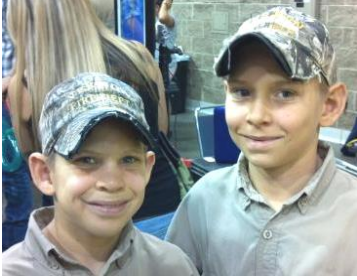
3. The study procedures include a health history and exam, a questionnaire, a urine sample (in some cases, also a stool sample) and a cheek swab for DNA (for the genetic analysis). The exam can be done by a healthcare provider in the local area. Travel to San Francisco is not necessary. The questionnaire will be administered by the study coordinator, over the phone. They will provide a kit for the sample collection, which can be mailed.

PLEASE NOTE: The Rare Disease Clinical Research Network, RDCRN, Patient Contact Registry is a method by which patients with rare diseases can register themselves with the RDCRN /Porphyria Research Consortium in order to be contacted in the future about clinical research opportunities and updates on the progress of the research projects. The APF will post an update on each of the projects and how to volunteer on the APF website, soon.

If You Can't Be A Research Volunteer, you can help by donating to research. There are six special research projects underway and all of them have very limited funding. Everything you give toward research goes to research. You can host a fund raising event, like Monica Fleegel who raised almost \$3000 at a fun dinner at her home. Or you can send a donation, big or small. You see, *the APF is YOU*. It is not an entity that has no connection to people with porphyria. Rather, *the APF is only US, all of US*. Research will affect US. We have this great opportunity to change the future for US and OUR CHILDREN. Every donation is important, so please do your part.

Porphyria Awareness Week provided the opportunity for all APF members to enhance *Porphyria Awareness* within their own communities and medical professionals. We appreciate the hundreds of our members who participated and thank them for their *Awareness* projects. Below are just a few stories of the APF members who hosted *Awareness* events—starting with kids and their parents.

Cook Brothers The Cook brothers, Cason (11) and Caul (10), have started a *Porphyria Awareness Week HAT DAY* tradition in their home town of Vernon, Texas. Both brothers have EPP and have set a great example on how to enhance porphyria awareness in their own local area. The boys wanted to raise money for the APF, too, so they asked the school board if they could do an awareness event and raise funds for the APF by hosting **HAT DAY** at



which everyone at their schools who wanted to participate could wear a hat in honor of the brothers and would bring in one dollar for the APF. The students loved the idea. It also gave Cason and Caul a chance to teach their schoolmates about porphyria, especially EPP. The boys explained about not being able to be in the sun and how it affects them, thus giving their friends a greater understanding of the severity and complexity of EPP. Those kids who had taken part in the event last year were eager to be a part of the event this year. They brought their dollars and wore their hats and made the event extra fun, educational and great for the APF. Also, employees at many

businesses could do the same by bringing a dollar and wearing a hat every day of *Porphyria Awareness Week*. Their whole community got involved and businesses even contributed by giving discounts to their customers. The Cook family also hosted another successful fundraising and awareness events at their church. This was such a wonderful and fun way to promote awareness and raise funds for physician education.

One of the most poignant notes was when the boys discovered a verse in the Bible they called their own, "*The Lord is like a shade tree at your right hand, the sun will not harm you by day nor the moon by night.*"

Alessia Callahan, who is a 5th grader from Greensboro, NJ, chose to make a presentation for her school as her *National Porphyria Awareness Week* contribution. Alessia has EPP and takes enlightening the public on the porphyria, particularly her type, EPP, very seriously.



Like the Cook brothers, she may be young but she has made a powerful impact on porphyria awareness in her community and school. She has educated her school for several years now by explaining the disease and sharing her own story with the students. To answer what EPP feels like, Alessia told her classmates, "*It feels hot - like there are needles going into my skin, and it takes a long time for that to go away.*" While Alessia and her family embrace *National Porphyria Awareness Week*

as an opportunity to share their story, their open approach continues throughout the year. "*Our family tries to get the word out as much as possible since her disease is not always visible,*" said Janeen, Alessia's mother. Both Janeen and Tim are diligent to spread the word about porphyria. We thank the entire Callahan family. Their local news printed an outstanding article about Alessia's effort and EPP. Read the article in the *Glassboro Schools'* news: <http://www.glassboroschools.us/site/default.aspx?PageType=3&ModuleInstanceID=767&ViewID=047E6BE3-6D87-4130-8424-D8E4E9ED6C2A&RenderLoc=0&FlexDataID=6698&PageID=1>

Pierre Mouledoux is also a longstanding APF member who has been very active in advancing EPP treatment and awareness. In fact, Pierre was one of the volunteers, who participated in the clinical trials to secure FDA approval for Afamelanotide, now named *SCENESSE*, which is an implant developed by Clinuvel Pharmaceutical. Pierre, like others, feels his time on the drug was the best, most pain free summer of his life. Pierre is so anxious to receive FDA approval of the drug that he has encouraged other EPP patients and his family to write the FDA on behalf of the treatment. Pierre has also volunteered to travel to Washington with Desiree/APF to represent EPP patients at a meeting with the FDA. Pierre will be sharing his own experience with EPP and how life was for him when he could finally have a normal life in the sun. Pierre has also



been a great encourager and friendly touch on Facebook among other people with EPP. He has also launched a campaign in Louisiana to confront their new laws that require fingerprinting and other rights that should not be violated just to get his windows tinted for EPP. Dave McCrae launched a similar campaign in the state of Washington to secure handicap parking when needed and won. We support and thank you, Pierre. *See you in Washington DC!*

EXCITING NEWS—APP FOR SAFE/UNSAFE DRUG LIST WILL BE AVAILABLE SOON!



Shawn Willis, an APF member (left with mask), has EPP. He has been very active both with the APF and the APF Facebook groups. Shawn has an amazing story to tell that shows us again that despite our health challenges, we can make a big impact in this world. Read Shawn's most recent adventure: *"Several months ago, I went to Uganda while in the last stages of the drug trial. I was so thankful to have had the active drug. In my opinion, at that time it was the only way I would be able to ever visit my friend, who runs an orphanage in Kampala. My trip was wonderful, and I thought that was the end of my travels until the drug finally comes to market. God had other plans. I began praying about going back again this spring. I was very nervous about being in Africa without the protection I had previously with the implant. I returned to Kampala in late March and I'm so thankful to say, everything went very well. While I was at the orphanage working outside, I would wear my mask. When we would travel in the car, I also wore it. Once we got to our destination, I would transition to a large brimmed hat I purchased from 'Sun Precautions.' I spent 10 days in Uganda and everything was wonderful! God is so faithful! If anyone would like to join me on a future mission trip, I would love to walk with you through the challenges of our condition while working to serve others who desperately need our help. The picture shows me all covered helping build picnic tables for the kids, because typically, the children eat on the floor of their homes."*



Monica Fleegel is a very busy lady as the Director of Human Resources at Mayo, but she gladly took on the heavy responsibility of traveling from Minnesota to clinical trials in Alabama for the Phase II and Phase III clinical trials testing SCENESSE. But she didn't stop there. Recently Monica gathered her friends and family for a triple fold purpose during Porphyria Awareness Week. She hosted a fantastic BBQ for family and friends as part of a fundraiser for the APF. While they were enjoying themselves, she also asked them to join in a letter writing campaign to the FDA to tell them about EPP

and Monica's experience from their viewpoint. They wrote about Monica and her struggle with EPP, how she could not enjoy normal daily activities like a simple walk outside or a trip to the beach. They also wrote about the greatest summer of her life when she was given the SCENESSE. Since the trials have a few months before completion and the data assessed, the attendees asked the FDA to approve the treatment quickly. Once again, without treatment, Monica and other EPP people are left to "fry" in the summer sun. The letters were all compelling. The Fleegel family has five siblings who have EPP, Monica, Theresa, Bill, Rita and Tom and all but one were on the clinical trials and all are in the Longitudinal Study. We sincerely appreciate the Fleegel family and all of Monica's family and friends who participated in this awareness and fundraising event. And thanks To Monica's daughter Addie, who made the APF cookie. **Yum!!!** Photo L to R Addie, Monica, sis-in-law Kathleen, brothers Pat and Tom.



APF Cookie

Victoria Harrold *"I am 28 years old and live in England. I was finally diagnosed with EPP at the age of 24 after a long struggle. Because of my own difficulty getting diagnosed, I now want to help raise awareness for this condition and encourage those who suffer in silence to fight to get someone to listen to them. My childhood in the spring/summer seasons was not a happy one. School was awful for me, because I was burning and no one believed me, and my skin was so scabby, the kids at school called me monkey, gremlin, alien etc. Even when my face was so swollen that my lip pushed up my nose and my knuckles disappeared when my hands were swollen and my feet were so swollen I needed a wheelchair, the doctors still continued to misdiagnose me. Doctors said I was attention seeking and put me on anti-depressants to cope. After years and years of misdiagnosis and hospitalizations, a new dermatologist who tested me for EPP solved the mystery."*



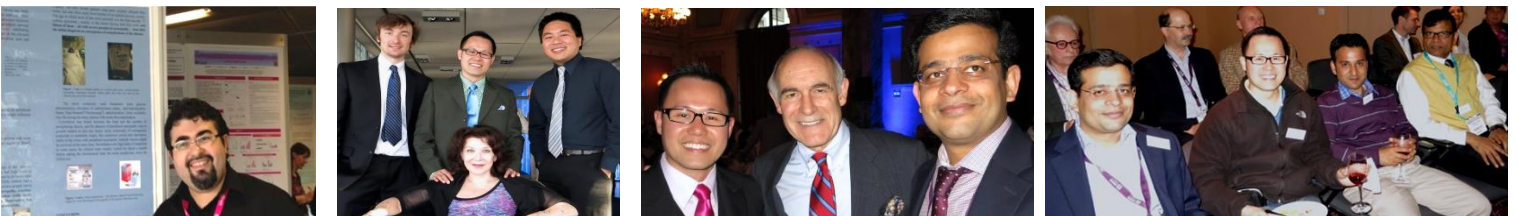
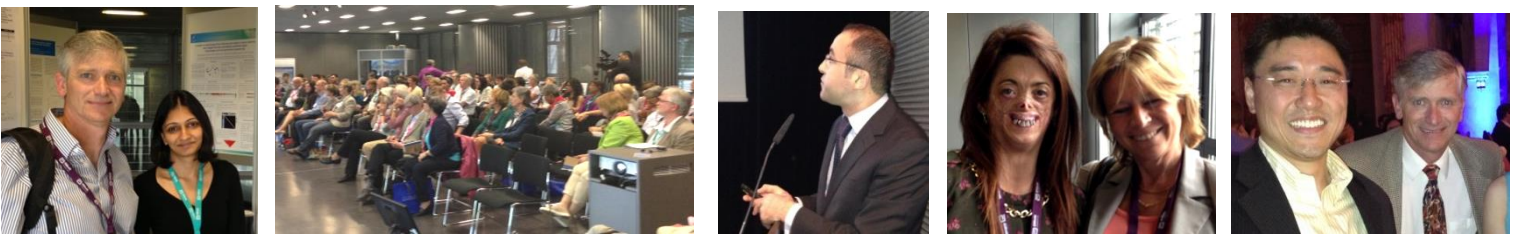
Jessica Hawkins Ivey helped with Awareness Week and our letter writing campaign to the FDA. Jessica participated in Phase II, the first round of clinical trials, and was very helpful to those who participated in the Phase III trials by sharing her experience. She also amassed a large number of letters for the FDA. Jessica has her Master's in Nursing and has worked in neurosurgery, orthopedics and nursing informatics. She now is the Nursing Clinical Manager in the perioperative division at a Midland, Texas hospital. Despite her busy schedule and long hours, she flew back and forth from her Midland home to the 'Galveston Porphyria Center' for the clinical trials. Jessica, thanks for all you have done for the APF and to move a new treatment forward for all EPP friends. *(Some of the above members have a full story on the APF website.)*

International Porphyrin Congress was a great success. Expert clinicians and researchers in the field of



porphyrins and porphyrias gathered from around the globe for this exciting meeting held every two years with the purpose of sharing their new research and clinical discoveries in a casual but private environment. To view the schedule, please see: <http://www.porphyrinsandporphyrias.org/> Although contributions from outstanding experts with an international reputation were featured on the scientific program, there was opportunity for all to discuss their newest discoveries, meet other experts and visit

colleagues from around the world. Many of our *Protect the Future* trainees attended the conference as well as made presentations. It was an outstanding opportunity for them to learn from internationally renowned experts and make friendships with other experts. The Congress also held **'Patient Day'** attended by patients from the USA, Switzerland, Germany, Romania, France, England, Brazil, Spain, Portugal, Sweden, Holland, Poland and Brazil enjoyed meeting one another, learning together and sharing their experiences. Thanks to Rocco Falchetto and Jasmine Barmine whose collaboration made **'Patient Day'** a great success. Both have EPP and understand the importance of patient collaboration nationally and internationally. They invited patients to join the online community, www.rareconnect.com, where patients can connect with patients around the globe. Photos **Row 1.** L to R, PTF Doctors and Mentors, (Drs Balwani, Ludtke, Latimer) (Larion, Yazici, Bonkovsky) (Singal, Bloomer) (Mittal, Singal) **Row 2.** (back~A.Lager, Perini, Gou, Caballes, front~Lager, Marcero, Mittal, Bloomer, Singal) (Gou, Anderson) (Gou) (Thapar, Yasuda, Balwani) **Row 3.** (Phillips, Naik) (Patient Day) (Yazici) (Fide-CEP, Sylve-French Foundation Exec Dir) (Wang, Phillips) **Row 4.** (Lourenco) (Larion, Caballes, Gou, Desiree) (Caballes, Desnick, Thapar) (front ~Thapar, Caballes, Mittal, Singal)



To read about the *International Porphyrins and Porphyrias Congress* see: <http://www.porphyrinsandporphyrias.org/> See *Experts' Patient Day* presentations : <https://www.rareconnect.org/en/community/porphyria/article/international-meeting-of-porphyria-patients-lucerne-2013>

Will A Hysterectomy Help ? Many women with acute porphyria attacks associated with menses want to know if having a hysterectomy will prevent acute attacks. Dr. Karl Anderson commented, "bilateral oophorectomy (removal of both ovaries) would be necessary to stop attacks. But it is usually better to use a GnRH analogue since it is reversible. If a GnRH analogue does not help, then taking out the ovaries will not help either. The uterus does not produce hormones, so hysterectomy alone would not affect porphyria. The uterus can cause pain if it contains fibroid, etc." *Watch the next newsletter for more information!*

RECORDATI Rare Diseases, Inc.



For the many patients who are taking Panhematin®, please note that Recordati Rare Diseases, Inc. has purchased Panhematin® from Lundbeck Pharmaceutical. Panhematin®, a treatment for the acute porphyrias, is the only commercial heme therapy available in the USA. Panhematin®, which is prescribed to correct heme deficiency in the liver and repress production of porphyrin precursors, almost always normalizes porphyrin and porphyrin precursor values. Recordati Rare Diseases, Inc. has assured us that there should be no interruption in receiving the Panhematin® treatment and that the product is the same as it has always been. In fact, for the next number of months, the telephone numbers and delivery services that were available through Lundbeck Pharmaceutical will remain the same, as well. Be assured that Recordati Rare Diseases is committed to porphyria patients.

FYI For questions from healthcare professionals regarding general use and administration of Panhematin®, please call *Recordati Rare Diseases*, **1.866.402.8520**. Your call will be answered by a member of the Lundbeck staff on behalf of Recordati Rare Diseases until late 2013. Your physician or hospital pharmacy can order Panhematin® through a wholesaler of their choice. *Healthcare professionals* can call **1.888.514.5204** to receive help with regular and emergency shipments, Monday thru Friday, 9 am to 6 pm EST. *After hours and emergency orders*, call **1.800.673.6723**. In an emergency event, when the hospital/physician/wholesaler does not have Panhematin® in stock, Recordati Rare Diseases will ship directly from its Laverge, TN distribution site via an expedited delivery service to anywhere in the U.S. — usually within 12-14 hours.

Panhematin® The APF has produced a new *Panhematin®* brochure. If you would like a copy of the new brochure for yourself or your physician, please contact the APF and we will forward one to you promptly. Remember, too, that the APF will send your doctor a free comprehensive doctor packet for the acute porphyrias. Call 1.866.APF.3635 or email porphyrus@aol.com. If your physicians have questions about the administration or use of Panhematin®, we will then send an instruction form or arrange a consultation with a porphyria expert, if needed.

'APF 2012 Presidential Award'



The APF presents its *Presidential Award* annually to the person who has devoted extraordinary service to the APF and our members. **Amy Chapman is our 2012 winner.** Amy's accomplishments and volunteer services are extraordinary beginning with the Blog she writes each week on the APF website, www.porphyrifoundation.com. Amy also serves as an administrator on the five APF FACEBOOK groups, answers at least 35 calls a week from patients around the country and shares her porphyria experience with each of them. If that is not enough already, Amy has taken on the leadership position of the international porphyria forum, www.RareConnect.com. Amy has made porphyria presentations to physicians and patients nationally and internationally, even on her vacations abroad. Many, many patients are wearing the purple porphyria bracelets and other APF memorabilia that Amy has sold as part of her personal fundraising effort to enhance our Physician Education projects. She has volunteered for research studies, serves on the APF Patient Advisory Committee, helped Desiree with special projects, hosted a patient support meeting, and has even been filmed in educational videos about porphyria. See: <http://www.youtube.com/watch?v=zxsnhxzBtyQ>.

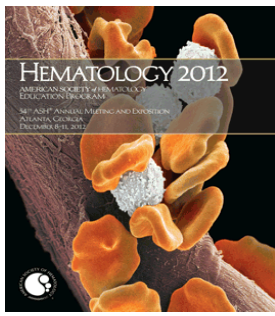
Amy and her husband, Craig, an IT specialist, live in the St. Petersburg area. Their recent move from Michigan was a big adjustment in climate and people and a great gain for patients in the area, because of Amy's wonderful ability as an educator and her willingness to provide a comforting shoulder. Amy is not all work and no play. She loves to travel, enjoys the water and beach and mostly being a volunteer not only for the APF but for a public ministry sharing her faith. We all owe Amy our sincerest gratitude. *Congratulations, Amy!!!!*

EPP Treatment News



SCENESSE®
Afamelanotide 16mg
Clinuvel's lead drug in development

The EPP trials are drawing to a close. Patients participating in the trials have one last research appointment before the trials end. The next step will be that all the data will be collected and assessed and sent to the FDA. Then the FDA will do its work and hopefully, approve SCENESSE/Afamelanotide as an EPP treatment. We have heard many, many wonderful stories of people who received the "real" drug and all of the fun they had at the beach, ballpark, fishing pond, family picnic, the daily events we take for granted. The excitement the participants shared about the success of SCENESSE was thrilling to hear. More exciting was that patients, who clearly received the placebo, continued to stay in the trials so that the research would not be compromised. All of the participants deserve our heartiest thanks. The APF will take one last step, too. We and trial participants will be meeting with the FDA to share their experiences both those who had the real drug and those who had the placebo. We also need people who were not on the trials but want to testify that there is a great need for treatment for EPP. If you live in the Washington DC area or live elsewhere and want to come with us to the FDA, please contact Desiree at the APF, 1.866.APF.3635 or email porphyrus@aol.com and we will call you.



Porphyria Publications Dr. Manisha Balwani, one of our *Protect the Future* (PTF) doctors, wrote an article with **Dr. Robert Desnick** that was selected by the **Blood and Hematology 2012 American Society of Hematology Education Program** editors for concurrent submission to **Blood and Hematology 2012**, namely, '**The porphyrias: advances in diagnosis and treatment**.' This is an outstanding article, <http://asheducationbook.hematologylibrary.org/content/2012/1/19.full> which may be of great interest to you.

The following articles are also excellent and very informative. Some PTF doctors are co-authors of the texts. Their names have been placed in bold. All of you who have contributed

to the PTF program should be very proud of our PTF doctors and the advancements they have made in porphyria.

* Anderson KE, Bishop DF, and Desnick RJ, (2001) in *The Metabolic and Molecular Basis of Inherited Disease, Disorders of heme biosynthesis: X-linked sideroblastic anemias and the porphyrias*, eds Scriver CR, Beaudet AL, Sly WS, et al. (McGraw-Hill, New York, NY), pp 2991–3062.

* Anderson KE, Bloomer JR, Bonkovsky HL, (2005) *Recommendations for the diagnosis and treatment of the acute porphyrias*. 'Annals of Internal Medicine' 142(6):439–450.

* Hasanoglu A, **Balwani M**, Kasapkara CS, (2011) *Harderoporphyria due to homozygosity for coproporphyrinogen oxidase missense mutation H327R*. *J Inherit Metab Dis* 34(1):225–231.

* **Yasuda M**, Bishop DF, Fowkes M, Cheng SH, and Desnick RJ (2010) *AAV8-mediated gene therapy prevents induced biochemical attacks of acute intermittent porphyria and improves neuromotor function*. *Mol Ther* 18(1):17–22.

* **Ryan Caballes F**, Sendi H, and Bonkovsky HL (2012) *Hepatitis C, porphyria cutanea tarda and liver iron: an update*. *Liver Int* 32(6):880–893

* **McGuire BM**, Bonkovsky HL, Carithers RL Jr., (2005) *Liver transplantation for erythropoietic protoporphyria liver disease*. *Liver Transpl* 11(12):1590–1596.

Porphyria Educational Materials The APF has a number of exceptional educational “must haves.”

1. We have a professionally produced DVD, **Porphyria Live**, which was filmed at two porphyria centers, Mount Sinai in NYC and University of Texas Medical School, with three experts, Dr. Sylvia Bottomley, Dr. Karl Anderson and Dr. Robert Desnick and a number of patients with each type of porphyria. Other interesting clips were interviews with caretakers, a clip of Karen Eubanks having her Panhematin® infusion and clips in the laboratories and clinical research units.
2. Desiree Lyon Howe's book, **A Lyon's Share of Trouble**, about her adventure as Executive Director of the APF and the thousands of patients she has met since the inception of the APF. Patients' own stories are included in the book, as well as detailed descriptions of each type and the many problems patients can incur.
3. The APF *Emergency Room Kit/Primary Care Physicians for Acute Porphyrias and EPP* are the most important material you can have in your educational arsenal. They include information pertinent to physicians and ERs.
4. A free very comprehensive Doctors packet for your own doctor.
5. A **'Warning Card'** for acute porphyria with the Safe/Unsafe Drug List web address noted.
6. Brochures like *Management of Acute Porphyrias, Panhematin®, Porphyria Cutanea Tarda, Diet and Nutrition*.
7. Materials for photosensitivity, like auto window film, sun screens, sun protective clothing.
8. **King George III** folder for the history buffs.
9. Testing instruction and fact sheets AND MANY, MANY, MORE!



In Memory We are saddened to hear of the passing of family members and friends.

Emmy Blume for *Kris Blume*; Lee Ann S Carson for *Terri Easterwood*; Mary P Crown for *Mary B Hargett Crown and Dean Puccia*; Elizabeth H Thompson, Patricia W Farnan, Susan Bardsley for *Sharon Charney Holtzinger*; Michael P Farina for *Vincent K Farina*; Kathleen Toelkes, Valine Jensen and Linda Peterson for *Donna Pagano*; Ralph M Gray for *Fred L Gray*; Norma Brown for *Matthew Brown*; Frank L Feczko for *Robert Zieles*; Stephanie Frazzini-Rushwin for *Suzette Frazzini*; Barbara Fielitz for *Patricia Woolard-Janes*.



In Honor We thank those who donated to the APF in honor of a friend or family members.

Jennifer R Ewing for *Desiree Lyon Howe*; Diana Parish for *Megan Parish*; Zila Reichman for *Tobey Reichman, Danielle, Arielle and Lielle Ovidia*; Robert P Quigley, Jr. for *Dr. Peter Tishler*; Lynne M Murray-Gray, Laurie Hanson, Ruth Wilson, Gary E Eyster, Grayfred B Gray, Mary Frances Donnelly, Larry Pritchard for *Ralph Gray*; Wayne R Whittenburg for *Jocelyn and Jamie Whittenburg*; Mary Ann M Kopie for *James and Robert McGuckin, Kathleen McGuckin, Georgia McGuckin*; Michael Pagano for *Andrea Pagano-Reyes*; Charlotte Beck for *Sarah Sundblom*; Richard J Drew for *Michael Drew*; Linda Nagin for *Melissa Nagin*; Anne T Wilson for *Candace Johnson*; Vasco L Walton for *Gaylene Parman*.



Exciting New AIP Treatment in Development Dr. William

Querbes, President of Alnylam Pharmaceuticals, made an exciting presentation at the International Congress about Alnylam's development of an entirely new class of innovative medicines based on a breakthrough discovery in biology known as

RNA interference, or RNAi. With RNAi technology, they have the opportunity to treat diseases and impact the lives of patients in a fundamentally new way by silencing disease-causing genes upstream of today's medicines. ALN-AS1 is a subcutaneous RNAi therapeutic targeting aminolevulinic acid synthase-1 (ALAS-1) for the treatment of acute intermittent porphyria (AIP). ALN-AS1 has the potential to be a therapy for the treatment of acute porphyria attacks, as well as a prophylactic approach for the prevention of recurrent attacks. *(Alnylam's name has interesting significance as Alnylam is the name of the center star of Orion's belt. The star has a luminosity that is 250,000 times greater than the sun, representative of the potential strength that RNAi therapeutics could bring to bear for human health.)* The APF will be announcing updates on the treatment and clinical trials.

APF Caretaker Support Group Warren Hudson, who serves on the APF Board of Directors, will head our new 'Caretaker Support Group' for spouses, partners or friends who help their loved one cope with porphyria.



This is a very important service because being a caretaker can be extremely difficult. If you would like to participate in the Caretaker group or speak with Warren, please contact the APF. Here is Warren's story. *"Porphyria is a large part of my life effecting relationships and day-to-day activities. It affects my ability to work, what kind of jobs I can take and how I take care of my family. Porphyria costs me friendships and takes a great mental and physical toll. I am extremely afraid of visiting new doctors, so I go prepared with questions and the expectation of disappointment or rejection. The horror related to a hospitalization is something I am unable to express. I know my story is common for those suffering from an Acute Porphyria. However, I do not have Porphyria. I am the husband and caregiver of someone with Acute Intermittent Porphyria (AIP). My experience lacks the physical pain but gives me a front row seat to hers and confronts me with an overall feeling of helplessness.*

The role of caregiver for a Porphyria patient has been largely unexplored. Obviously, the patient is everyone's primary concern—as they should be our focus. However, a strong healthy caregiver provides better long-term support. It is all too easy for both caregiver and patient to focus on the patient. In my case, this comes with consequences. At first, both my wife and I treated AIP as something separate from our relationship and our lives. This was unrealistic and fed many disappointments for both of us when we realized the disorder could not be ignored. We strive to understand how it is part of our lives and work together to limit its effect. I wanted to share some experiences, provide insight and hopefully help others in my position. Porphyria caregivers also rarely communicate enough with other caregivers and often bottle up their emotions, focus solely on our loved ones and in my case, internalize my frustrations and fears. This can be isolating and unhealthy. I took my personal concerns, pains or illnesses and measured them up against what my wife experienced. In comparison, my problems seemed petty. If I got sick, I treated my illness like something I should push through or shrug off. Not caring for myself, affected my health resulting in a large weight gain and depression. Even when those closest to me were concerned and wanted to talk, I held back details because it felt like I was betraying my wife by discussing her AIP and how it affected me.

The irony of putting someone else's needs ahead of your own is that both people suffer. It is critical that those of us in the caregiver role take the time to ensure our own needs are being met and that we do not neglect the most important person, ourselves. In order for our spouses and loved ones to get the care they need, it is imperative that we take care of ourselves both mentally and physically. Reach out to other caregivers if you need advice or a sympathetic ear. Get involved in things that bring you joy. These have been hard lessons for me to learn, and something I struggle with on a daily basis. If you remember anything else, please know neither the patient nor the caregiver is alone in this journey. **See the entire article on the APF website in the new 'Caretaker Section.'**

'Warning Card' and Doctor Packet If you do not have a free **APF Wallet 'Warning Card'** for acute porphyria or a free Doctor Packet for your physician, please request one or both from the APF by emailing: porphyrus@aol.com or calling 866.APF.3635. The cards are a perfect fit for your wallet and include the URL for the **Safe/Unsafe Drug List**.

The **Doctor's Packet** is very comprehensive and includes, CME course information, drug information, *Pan-hematin* brochure, *Management of Acute Porphyrias* brochure, as well as major medical journal articles written by our Scientific Advisory Board of experts. Over 3000 doctors have either requested this packet or received them from patients. This packet educates your doctor how to diagnose and treat the acute porphyrias. Please send the APF your doctor's contact information, and we will mail a packet to them immediately.