



**The American Porphyria Foundation is your foundation.**

Thank you for your generosity, which enabled us to better your health by enhancing public awareness

about porphyria, educating patients and physicians about porphyria, and supporting research to improve treatment and the search for a cure.

**Yvette, Africa, Carol, Sarah, Mira, April and Desiree**



**Merry Christmas and Happy Holidays!**



*Photo — Top Row: Africa Hernandez, Terri Paynter, Abril Hernandez, Yvette Strange, Maria Gonzales — Seated: Desiree Lyon, Judy Pereira*

**NIH Gives \$4.4 Million for Porphyria Research**

We are thrilled to announce that the National Institutes of Health has awarded a consortium of porphyria experts a \$4.4 million grant to study these diseases. The Congress also notified NIH via legislation that porphyria should be a national research priority (see p. 7). Who could ask for a better present than that?

This means the doctors can build a registry dedicated to the study of porphyria treatment and begin several important clinical studies in acute porphyrias, PCT and EPP. We will finally be able to answer the questions: "How many people in the United States have porphyria, and which kind do they have?" But not without your help!

With this grant NIH recognizes that the porphyrias are grave diseases that affect an active and involved community of patients, families and physicians. APF member participation was key in this. The grantors wanted to know that people with porphyria and their families would write letters, visit their congressmen and contribute. That is why we came to you last year with a challenge grant. You came through, giving \$100,000 to this effort and earning us a matching grant of \$100,000 per year for five years from an anonymous donor. That combined gift convinced NIH that porphyria research is crucial to all of us.

Together, we have made more research possible.

This grant is an imperative to build a lasting network for porphyria research in the United States. It will be used to build a network of five clinical research centers across the United States. Let's not

waste this momentum! We need to take action *now* to sustain and build these projects beyond the current grant period and for the rest of *our* lives.

Competition for this grant was fierce and the consortium members' work was selected to receive this prestigious award from among many excellent proposals. Please join us in thanking the following doctors especially for their monumental efforts on our behalf: Dr. Karl Anderson, Dr. Robert Desnick, Dr. Joseph Bloomer, Dr. Montgomery Bissell and Dr. James Kushner.

**We have two big tasks ahead:**

**First**, we need to locate porphyria patients who are not already in touch with the APF so that they can be part of the registry. With their permission, please let us know the names and contact information of all in your household and family who have porphyria. This will enable the doctors to contact them for inclusion in the registry.

**Second**, we must all do some creative thinking about how we can raise funds to **Protect the Future** by training the next generation of porphyria experts and supporting porphyria research. The APF is all of us, because the APF serves all of us. We beseech each and every one of you to think about how you can contribute to building an infrastructure for porphyria education and research—for our own lives and our children's.

— **Desiree Lyon Howe**



## **New Health Institutes Director: A Rare Disease Advocate**

Francis S. Collins, M.D., Ph.D., took his post this summer as Director of the National Institutes of Health (NIH). Dr. Collins formerly directed the National Human Genome Research project that mapped the human genome, which he called "the book of human life."

The APF is particularly excited about this appointment because Dr. Collins has been an ardent supporter of rare disease research, noting that understanding and developing new treatments for rare diseases will be key to understanding and treating more common conditions.

In a 2000 White House ceremony marking the release of the genome's first draft, Dr. Collins said: "It is humbling for me, and awe-inspiring, to realize that we have caught the first glimpse of our own instruction book, previously known only to God." The NHGRI project culminated with the sequencing of the entire human genome in 2003. Dr. Collins is also known for emphasizing the importance of addressing legal and ethical issues in genetics.

Dr. Collins was awarded the Presidential Medal of Freedom for his work on the NHGRI. In October of this year, he received the National Medal of Science. Dr. Collins's laboratory also discovered the genes responsible for cystic fibrosis, neurofibromatosis, Huntington's Disease and others.

Dr. Collins is famously interested in the coexistence of science and faith, and has written about this in *The Language of God: a Scientist Presents Evidence for Belief* (2006). His new book, *The Language of Life: DNA and the Revolution in Personalized Medicine* is to be published early next year. He has also participated in efforts to popularize science, recently appearing in a photo shoot for the Geoffrey Beene ad campaign *Rock Stars of Science*.

## **Dr. Geoffrey Dean, 1918-2009**

We are saddened by the death in Dublin, Ireland of Dr. Geoffrey Dean, a noted porphyria expert who made significant contributions to the genetics of Variegate Porphyria. Dr. Dean's contributions to medical literature continued until the end of his life, with a chapter on porphyria and epilepsy he recently completed to be published soon.

Born in England, Dr. Dean spent the years 1947-1968 working as a private and state physician in South Africa. He was working in and near Port Elizabeth when he learned about the case of a nurse who was ill in Cape Town's Groote Schuur Hospital, now the site of one of the world's best known porphyria centers. Undiagnosed, this young woman was given a barbiturate, an unsafe drug that caused her death. The patient's father told Dr. Dean that he and other members of the family had the same undiagnosed sickness.

After the father was diagnosed, Dr. Dean traveled the country with a nurse. They collected blood samples from families who reported a similar illness, laying the groundwork for a 118-family genealogical study and identifying Variegate Porphyria. The great Lenox Eales later took over this work and put South Africa on the map medically and genetically. Although quite rare everywhere else in the world, VP is the most common genetic disease in South Africa, found primarily among the Dutch-descended (Afrikaner) population.

The prevalence of VP in South Africa is due to something called the "founder effect." In this case, a young Dutch immigrant couple, one of whom had a mutation for VP, married in 1688. The small size and isolation of this group created perfect conditions for distributing the VP mutation widely in the population.

Dr. Dean's work eventually enabled scientists at the Porphyria Centre in Cape Town to confirm that the R59W mutation most South African VP patients share originated in the Netherlands.

### **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 porphyria. Please join us in thanking: Geraldine and Jackson Williams for Birtie M. Prine, Carole E. Kuklewski for Vince Kuklewski, Alda Davis, Carolyn Blue, Linda E. Armi for Lori Woods, Dolores M. Brazas for Wesley J. Brazas, B.B. Ferguson, Carol L. Smith for Norma Lambert, Marianne and Arthur Danko for James Smuczynski, Ellen Joyce for Fagie (Kubersky) Toback, Chelsea and Lisa Kancsar for Stephen Kancsar, Cantigny Research Foundation, Inc. for Dr. Richard D. Levere, Frances Mauro for Louis J. Mauro.*

### **In Honor**

*Others have chosen to honor their friends through a gift to the APF: Charlotte Beck for Elva Denger, Joan Steelhammer for Jann Steelhammer, Robert E. Christensen for Richard Dugger.*

*The American Porphyria Foundation disseminates accurate medical information to patients, educates physicians in appropriate diagnostics and care for the porphyrias, and supports 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.*



## Liver Group Honors Dr. Bloomer

The APF is proud to announce that our own Dr. Joseph Bloomer was honored at this year's meeting of the American Association for the Study of Liver Diseases with the organization's Distinguished Service Award. Dr. Bloomer is a founding and continuing member of the APF Scientific Advisory Board and has contributed enormously to study and treatment of the porphyrias, especially the acute porphyrias and EPP.

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The AASLD Distinguished Service Award is given to honor sustained service to the organization or to the liver disease community over an extended period.

Dr. Bloomer is a past president of AASLD and expert in transplant hepatology and EPP liver transplant. He has served for several years as a mentor for young doctors in the APF **Protect the Future** program, working closely with Dr. Brendan McGuire, now a porphyria specialist in his own right and a transplant hepatologist together with Dr. Bloomer at the University of Alabama-Birmingham (UAB).

Dr. Bloomer leads porphyria work at UAB, and has received NIH grants to help support his porphyria research for more than 30 years, with significant contributions to the understanding of porphyria genetics during that time. UAB now houses a porphyria study center, under Dr. Bloomer's leadership, as part of the new NIH grant for porphyria (see p. 1). As an EPP specialist, Dr. Bloomer will study Afamelanotide in EPP patients (see p. 5).

Congratulations Dr. Bloomer, we're so proud of you!

## APF Exhibits at Liver Meeting

The APF was joined by many eager volunteers at this year's meeting of the AASLD, the major professional organization for hepatologists in the United States. Members came to Boston from the surrounding area to educate physicians about diagnosis and treatment of the porphyrias by sharing their own experiences and the APF's materials. The APF has built a reputation as a reliable source of medical information over several decades, and physicians seek out our literature and services to assist in caring for their porphyria patients.

All of the medical literature we distribute is written and approved by members of our Scientific Advisory Board, each of them an expert with a 30-year record of conducting porphyria research. We adhere to this standard because it is vital that patients and their doctors have a reliable place to turn for guidance when dealing with the porphyrias, which can be confusing and frightening diseases.

Thank you to all the volunteers who came out to staff the booth at AASLD: Patricia Weiss, Charles Johnson, Sarah Litchfield, Michael Drew, and Terri Kitcoff. Your contribution was tremendous and we couldn't have done it without you!

## Protecting Our Future: Dr. Tarun Narang



One program not covered by the grant from NIH (see p. 1) is the APF **Protect the Future** program. This means we still need your help to insure that we will all have doctors who make a serious study of porphyria to care for us and advise our physicians in the future.

This initiative has so far been quite successful in attracting young specialist physicians to train as part of the next generation of porphyria experts. With your help, physician-researchers with many decades of specializing in porphyria have trained 10 new doctors in treatment and study of the porphyrias. Now Dr. Herbert Bonkovsky is training Dr. Tarun Narang at the Carolinas Medical Center in Charlotte, North Carolina.

Dr. Narang is a fellow in transplant hepatology and completed his medical residency in Internal Medicine at the Weill Medical College, Cornell University in New York. He won the annual Hippocrates Awards Research Competition for 2009, and is fluent in English, Spanish, Hindi, Urdu and Punjabi—a plus for a porphyria expert, as specialists in the United States receive calls from desperate porphyria patients all over the world.

We welcome Dr. Narang and look forward to many years of working together!

## Acute Porphyria & Birth Control

Hormones can be tricky for women with acute porphyria. While some women with acute porphyria can tolerate oral contraceptives ("the pill"), the medication is listed on the **Unsafe Drug List for Acute Porphyrias** and can cause an acute attack. Experts advise patients to avoid medications thought to be unsafe in acute porphyria if at all possible.

Many birth control pills contain progestogen, which is particularly precarious for a woman with an acute porphyria. The "morning after pill" also contains progestogen and can precipitate an attack.

Certain intra-uterine devices (IUD) contain progestogen. Because the progestogen in the IUD is localized in the uterus, it enters the bloodstream incrementally. This type of IUD has shown little risk of causing an attack. The copper coil (IUD) is hormone free, as are condoms and other "barrier methods" of contraception. Please consult your physician about hormone-free options for pregnancy prevention and all other medical questions!

## Hormones & PCT

Estrogen has been shown to activate Porphyria Cutanea Tarda (PCT), and doctors may advise patients to stop taking it while they suppress a flare-up of the disease. Estrogen is found in oral contraceptives and is sometimes used to treat prostate cancer. People with PCT should discuss medication choices with their doctors, but note that PCT is **NOT** an acute porphyria, so the safe and unsafe drug lists for acute porphyrias do **NOT** apply to patients with PCT.

## 2009 APF President's Award Dr. Karl Anderson



Like most rare disease experts, Dr. Karl Anderson, chairman of the APF Scientific Advisory Board, is consulted by patients and physicians from around the world. Such calls are time intensive because porphyria is a complicated disease and is unfamiliar to most doctors. Yet Dr. Anderson helps these people because he understands that patients with porphyria often have nowhere else to turn.

At the APF, we hear wonderful stories about Dr. Anderson's compassion and willingness to help very sick patients. Dr. Anderson treats patients respectfully and demonstrates great understanding of the complex medical maze so many wander on their path to correct diagnosis and proper treatment.

Dr. Anderson has been internationally renowned in the field of porphyria for three decades. He serves as Professor in the Departments of Preventive Medicine and Community Health, Internal Medicine, Pharmacology and Toxicology; Director of the General Clinical Research Center; and Director of the Porphyria Center and Laboratory at the University of Texas Medical Branch in Galveston, Texas. He has published widely on the porphyrias, both in major medical textbooks and medical journals. His clinical research on the porphyrias has included heme therapy, porphozym and LHRH for the acute porphyrias, characterization of the enzyme defects in the different forms of porphyria, and susceptibility factors and treatment options for porphyria cutanea tarda (PCT). His work has been instrumental in the development of the current standard of care for porphyria and basic scientific and medical understanding of the disease. Many of the APF **Protect the Future** clinicians have trained in Dr. Anderson's clinic and laboratory.

Dr. Anderson is a quiet, humble gentleman, as esteemed among his peers as he is among his patients. We are proud to have worked with Dr. Anderson for nearly 30 years. His contribution to understanding and awareness of the porphyrias has been exceptional and it is our great pleasure and privilege to give him the APF President's Award for 2009.

### Let's Get Back in Touch!

This issue of the newsletter is going out to many members we haven't heard from in a long time. We are reaching out now because we have so much exciting news to share with you and we want you to benefit from current research. But the APF has been hit hard by the economy and we cannot often afford to send newsletters to members who are not up to date. If you've been out of touch, won't you please call our office to renew your membership or use the enclosed envelope to send in your \$35 annual donation? We depend on you; we depend on each other. Thank you!

## APF President's Award James and Deb True

The APF President's Award is given to a member for outstanding contribution to porphyria awareness, advocacy and research. James and Debra True made such a contribution this year when James completed the Coeur d'Alene Ironman Triathlon in June and the family raised \$4,000 for the APF from friends and loved ones who donated money to express their support. The *Janus Charity Challenge* matched the funds the True family raised, doubling their contribution to us!



James and Deb have struggled for years with Debra's bouts of porphyria. Because of Debra's illness, the family wanted to raise funds for the APF. So they shared their story with everyone they know and solicited funds to help fight porphyria.

Through their fundraising work, James and Deb have not only supported the APF's programs, but educated their local community about porphyria as well. And as anyone who has porphyria knows, awareness is our first and biggest hurdle. Education is the first step in improving our diagnostics and treatment, and eventually finding our cure.

Please visit our website to read James & Debra's story: <http://www.porphyrifoundation.com/about-the-apf/member-stories/deb-and-james-true>



### Getting Ready for the Holidays?

#### Benefit Porphyria Research While You Shop!

Just in time for the holiday season, the following vendors have agreed to help you support the American Porphyria Foundation:

*Make Someone's Day Today* is a line of small, affordable gift baskets for all occasions. A division of The Basket Lady, *Make Someone's Day Today* donates a portion of every sale to the APF.

Upscale Resale Consignment Boutique helps you turn upscale women's clothing you no longer wear into a donation to the APF. Call our office at 1-866-APF-3635 for details.

Amazon.com will donate a portion of sales when you shop via this special web address:  
[http://www.amazon.com/?\\_encoding=UTF8&tag=ameriporphfou-20](http://www.amazon.com/?_encoding=UTF8&tag=ameriporphfou-20)

Please see the APF website Products and Publications page for more details and to connect:  
<http://www.porphyrifoundation.com/get-involved/publications-and-products>

And don't forget, you can always choose to honor a friend or loved one by making a gift to the APF *In Honor* or *In Memory* of someone special. We thank you.

## The Leppert Family: EPP on Dr. Oz



Show in October to raise awareness of EPP: what the disease is, its symptoms, and its effects on the lives of two otherwise healthy teenagers.

Even though the Leppert family has known how to deal with EPP for years, mom Tracy says there are still difficult times and lonely times, for instance when one of the children's friends invites them to a pool party in the middle of the afternoon. Tracy emphasized that her children can still do anything a child without EPP can do, they just have to do it at night. "We have pool parties too," she says, "until two in the morning!" Nicole told Dr. Oz that her favorite after-hours event so far has been water-skiing in the moonlight during the family's annual trips to Maine.

Craig pointed out that his family has adapted to life with EPP, and being able to achieve all he has despite having EPP has been "that much more rewarding," but he still "wouldn't wish the kind of pain that I or my sister Nicole have felt on anyone." Craig's goal is to achieve enough that others will think of him as a three-year starter on his high school's varsity football team and senior-year President of the school student body, rather than defining him by his illness.

In the studio audience was Dr. Lisa Sanders, who writes a monthly column in the *New York Times* about a difficult-to-diagnose case. Toward the end of the segment, she gave valuable advice about how patients with rare or difficult to diagnose diseases can participate in their own diagnosis. Dr. Sanders encouraged patients who think their diagnosis may be incorrect to discuss this with their doctor, and to ask her or him about getting a second opinion if it seems your doctor really does not know how to help you. (Dr. Sanders's November 1 column featured a case of acute porphyria.)

Dr. Oz further suggested that when you research your symptoms online, you seek out articles in medical journals that are written by MDs and take note of who is writing those articles. Those are the experts you or your doctor will want to seek out for help finding the right diagnosis.

*I'm So Excited for EPP Trials!* — Today I received a form for the EPP clinical trials. You cannot imagine how excited I am! It would be a dream come true for me to be able to participate in these trials. Doctors used to tell my mother just to keep me out of the sun and that there was nothing wrong with me. I am 65 years old and have lived with EPP as far back as I can remember. That hot burning sun, day after day. I was almost 40 years old when I was diagnosed. I had driven eight hours in the daylight and was in excruciating pain. A doctor finally gave me a diagnosis, but no relief. I stayed out of the sun when I was young and when my children were young. Now I finally have a chance of being in the sun with my grandchildren. **I have waited 60 years for some kind of help.** My hopes are very high for these clinical trials! Thank you APF. — **Sandra Bolding**

## Monica Foley Fleegel: EPP in a Big Family

My name is Monica Foley Fleegel, and I come from a family of 10 kids. Five of us have lived with EPP our whole lives. Having the disease was very difficult for all of us, including the siblings without the disease, because all the family's plans were made around those of us with EPP. However, it shaped us into the adults we are today. We all chose careers that would allow the least amount of time in the sun and we all have hobbies and activities centered around being indoors.

We are now all in our late-40s to mid-60s and those of us with EPP have figured out how to live our lives out of the sun. But as we age, have children and now grandchildren, it is difficult not to be able to join in with their activities. I know we all still take risks and then suffer the pain of an EPP reaction—which usually makes it not worth the experience!

I dream about the day I might be able to walk to my car after work without pain or go with my daughter to the amusement parks she loves so much and not have to watch her day on videotape.



### EPP Trials News

After joining several EPP specialists at meetings with the FDA Office of Orphan Products Development, we are more hopeful than ever that trials for **Afamelanotide**, the potential new medication for Erythropoietic Protoporphyrin (EPP), will begin next spring! Now our task is to identify potential research volunteers. It is vital that volunteers identify themselves and family members with porphyria who would like to be included. That way the researchers and FDA will know how much interest there is in finding new treatments.

Trials have been in progress for several years already in Europe, with very promising results. The medication is a little pellet about the size of a grain of rice, which is implanted under the skin. The pellet dissolves as the medication is absorbed, and when it is completely absorbed, a new dose is given. Patients have been so happy with the results that they've asked to be allowed to keep taking **Afamelanotide**.

Even if you've contacted us already, if you're not sure you want to participate in a trial, or if you have more questions for the doctors, please call or write to us. Be sure to give us the name and contact information of everyone in your family who has EPP and wants to be included.

## Dr. Shedlofsky Speaks At OH Meeting



A group of about 20 patients, family members and supporters recently met at the home of APF member Steve Stevens to share experiences, visit, and hear Dr. Steven Shedlofsky give a presentation on the porphyrias. Dr. Shedlofsky has devoted more than 25 years of his medical career to researching and treating the porphyrias, PCT and the acute porphyrias in particular, and serves on the APF Scientific Advisory Board.

Meeting attendees were deeply grateful to Steve for making his home available and organizing the event, and to Dr. Shedlofsky for sharing part of a Saturday to educate them about porphyria. Enthusiasm for the meeting was evident in the distance participants drove to be there—some coming from as far as four hours away on a Saturday afternoon.

Dr. Shedlofsky's presentation touched on the differences among the different types of porphyrias—the cutaneous porphyrias (PCT, EPP, CEP) that cause the skin to react painfully to sunlight, and the acute porphyrias (AIP, HCP, VP, ADP) that cause episodic attacks of severe pain and a variety of other symptoms. Dr. Shedlofsky described the chemical processes in the body that cause porphyria symptoms, the difficulty of diagnosing these diseases and the proper approach to diagnosis, and finally the means of treating the different porphyrias.

Being diagnosed with a form of porphyria can be alarming because all of these diseases are so little known and so much of the information available is inaccurate or conflicting. Dr. Shedlofsky has pointed out the absolute necessity of relying on well-regarded sources of medical information when you do research on the Internet, like the National Institutes of Health, Centers for Disease Control, Food & Drug Administration or American Porphyria Foundation. For those not sure where to turn for information, try the APF website's **Additional Resources** section, which includes links to medical websites and reliable porphyria websites in other countries. The most important thing for newly diagnosed patients to know is that whatever type of porphyria you have, there is treatment available. Please contact us, or have your doctor contact our office for more information. We are here to help you.



## Thank You Terri!

Has anyone ever entered your life at the perfect moment? That is how we felt when Terri Paynter volunteered to help us at the APF office. With two major medical conventions coming up, plus physician training programs and the added work of supporting the national porphyria registry and new clinical studies. We were on overload and in great need of help. Terri arrived just in time.



Terri had been very sick and often in the hospital with acute porphyria. When she returned home from her last hospitalization, she reached out to Nita Busby, who also has porphyria. One day, they began talking about God's purpose for their lives. Nita asked Terri: "what is your purpose in life?" Shortly after, Terri called Desiree at the APF office and asked if she could volunteer to help.

Terri is smart and fun, and has been a GREAT help. She is a wonderful asset and a blessed new friend. As you can see from the photo, Terri is willing to do anything and work anywhere, including the office floor! *Thank you Terri.* Your purpose is to educate people about porphyria!

*Read about our newest volunteer in the March issue. If you live in the Houston area and have some time to spare for the APF, there's always extra work for a willing pair of hands. 1-866-APF-3635 for details.*

## Life-Saving Information

When it comes to porphyria, "those who know the most do the best!" So we disseminate accurate medical information directly from porphyria specialists. Together we have moved mountains to advance porphyria research on the largest scale ever in this country. We could never have done this without you! But this economy is terrible for small organizations like the APF.

The Canadian Porphyria Foundation was forced to close its doors this year for lack of funding. Thankfully, we are still afloat, but we need your full participation to stay that way. Because of education and awareness campaigns, and the spread of information on the Internet, our office receives more requests for information than ever. The APF also facilitates consultations between local treating physicians and porphyria experts—often in the midst of a medical crisis. Providing these services for those in need is extremely gratifying. But meeting all these requests is also expensive.

So we turn to you. If your annual membership of \$35 is up to date—especially if you have chosen to donate above and beyond \$35—thank you so much. Give yourself a pat on the back because YOU are making life-saving information available both for yourself and for others. If you have not donated in a while or are unsure of your membership status, please take a moment to consider what the APF means to you, and what you can give, and then call us today.

We are here for you. We exist to serve our members and people with porphyria. *Thank you* for your support.

## AIP in NZ: Kim Willis-Bregmen



My wife Kim and I live in Otaki, a small rural community in New Zealand. Kim was admitted to our nearest hospital in 2003 with abdomen and back pain, vo-miting and sore arms and legs. Her blood pressure rose dangerously high, and she suffered a mini-stroke and multiple seizures and

spent days in intensive care whilst tests were done to investigate the diagnosis of porphyria. Kim's mother and grandfather both died of a condition that fit the symptoms of porphyria, but were never diagnosed. In Kim's case, tests were completed and AIP was the diagnosis.

Kim spent 13 weeks in hospital and rehabilitation during this first visit. Her need for physiotherapy was a result of the major nerve damage she suffered. 30 times over the next three years, Kim would be admitted through the Emergency Department, then receive delayed treatment or the wrong treatment, making her condition worse.

Eventually I began searching the Internet and joined the American Porphyria Foundation. I got information about managing this condition and what treatment was used overseas. Without this support we might still be struggling to this day.

Kim was finally treated with heme therapy for the first time in 2005 with a marked improvement in pain control and reduced time spent in hospital. Eventually we got to a position where Kim received heme therapy early in an attack and this stopped the attack before it progressed and did serious damage. Kim's story is really about the struggle of living with AIP for years with no support network until our lucky break with heme therapy.

Kim has ongoing issues with abdominal pain, back pain and pain in her limbs, which she manages with a better diet and the removal of all things that could precipitate another attack. Kim takes medication for pain and sometimes takes a few days out of her busy job. All things considered, she now lives a pretty normal life.

We are confident that with the knowledge, support networks and treatments we now have in place, we are equipped to tackle this purple monster head-on should we ever have the misfortune to encounter its fury again. — **Blair Bregmen**

## New Friends in Switzerland

The American Porphyria Foundation was very happy to receive the following introductory message from our newest Global Partner, the Swiss Porphyria Association: *The Swiss Porphyria Association was founded on January 31 of 2009, so it is still a very young, small organization, operated entirely on a volunteer basis. We are preparing our first official General Assembly, for January 2010. We are looking forward to working towards strengthening and growing our group for the benefit of Swiss patients.* <http://www.porphyr.ch>

Please join us in wishing our new friends in Switzerland every success as we work together to improve research and care for porphyria patients in both our countries!

## Carol Coons: My Road to PCT



I have PCT but was misdiagnosed many times since I first noted that I was blistering in 2004. Three dermatologists in a row were unable to figure out what was wrong with me even after I had a biopsy. My ferritin level was accidentally discovered at an unrelated clinic visit—it was over 900.

At that time, I was told to go to a hematologist, who said that my problem was hemochromatosis, even after I showed him my blisters and sores. I did have a genetic H63D heterozygous defect. I had a port put in place and to date, I have had over 45 phlebotomies. My internist sent me to get a second opinion, where I was told that I had PCT. As the treatment was the same, I had not gotten a port or had the other phlebotomies in vain.

I have no idea how I developed PCT, but now I try to stay out of the sun. The PCT showed up while I was taking Paxil for my fibromyalgia pain and spending a week at the beach. Now I try to stay out of the sun and am careful not to eat too much iron. My liver is fine now, and fortunately I did not need a liver biopsy.

*Editor's Note: Although PCT is the most common type of porphyria, it is often misdiagnosed and under-treated. The APF is looking for young doctors to join the **Protect the Future** program with an eye to becoming PCT experts. Please consider making a donation to the APF to support this training, and note that it is for **Protect the Future/PCT**.*

## Congress to NIH: Porphyria is a Research Priority!

The U.S. House and Senate have instructed the NIH Office of Rare Diseases Research to "develop an agenda for basic and clinical research for the treatment of porphyria, to devote dedicated resources for this purpose and to consult with patient stakeholder organizations when considering the development of the research agenda."

Porphyria is one of only a handful of rare diseases to be named in a congressional message to NIH. This funding opportunity occurred because of the efforts of many friends, especially Camp Kaufman and his colleagues at Cornerstone Governmental Affairs, who donated their services. Without their work on our behalf, we would not have received this coveted award. Special mention is also due to Kasirer Consulting and APF member Edward Geffner for their invaluable assistance. Other members were essential links in the chain with letters to their representatives and senators.

We still need your help with future efforts! We are seeking committee members with PCT and EPP for the Media Relations and Government Relations Committees. The time you volunteer to the APF really does make a difference in your future! Please consider participating. — **Desiree Lyon Howe**



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*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](http://www.porphyrifoundation.com)

**Doctors are Recruiting Patients Now** for clinical research studies. A \$4.4 million grant from NIH has jump-started clinical studies for PCT, EPP and acute porphyrias. Please call our office for details or to be contacted by researchers.

**Holiday Gift Giving:** Remember the APF in your holiday shopping this year. Visit Amazon or other vendors via the APF website, or honor a loved one with a donation. See page 4, or call our office for details.

Tell your doctor about the **Safe/Unsafe Drug Database for Acute Porphyria** and **Emergency Room Guidelines for Acute Porphyrias**. All medical information we distribute is written by porphyria specialists.

**Is Your Membership Up to Date?** Don't miss a newsletter! 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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