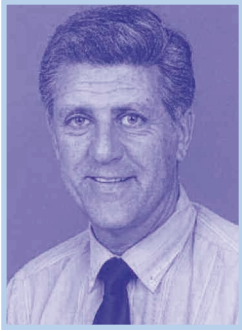




Educating to Save Lives — National Porphyria Awareness Week, 2009 **Raising Awareness to Save Lives: Highlights from National Porphyria Awareness Week**

Sound(ing) Off! About Porphyria on KPRL



Dr. Neville Pimstone

Monica Firchow returned this year to *Sound Off!*, a popular mid-day talk radio show on the California Central Coast, with host Dick Mason. Mr. Mason invited Monica to appear on the show with porphyria specialist Dr. Neville Pimstone and the APF's Mira Geffner to help educate the public about porphyria and discuss her forthcoming biography of her father.

Monica spoke with Mr. Mason about her father, Gene Bennett, who fought mightily to live a normal life with Congenital

Erythropoietic Porphyria (CEP), an extremely rare form of the disease that causes severe skin damage and anemia.

Dr. Neville Pimstone, a member of the APF Scientific Advisory Board since our founding, contributed information on the science of porphyria and spoke movingly about his relationship with Mr. Bennett, whom he treated for many years. Mira discussed the APF's work, the importance of porphyria awareness, and her own symptoms with AIP. We are grateful to Mr. Mason for providing such an open and welcoming forum for this discussion.

For more information on Gene Bennett, see Monica's website: www.genebennett.net

Lisa Kancsar, NPAW Fundraiser

As she does every year, APF member Lisa Kancsar commemorated National Porphyria Awareness Week by raising more than \$700 at the hospital where she works as a nurse. Lisa has always been there when we need her — educating the hospital staff and raising money for vital research and education projects.

Thank you Lisa!

Megan Railling on National Radio

APF members will recognize Megan Railling's name as the young woman who first became ill with Acute Intermittent Porphyria (AIP) while still in high school, but was not diagnosed until she was halfway through college. Megan's first frightening symptoms are familiar to acute porphyria patients: acute vomiting and loss of bowel function, terrible pain and neuropathy, and a dramatic weight loss caused by her inability to eat.

Megan's health has improved dramatically since her diagnosis. She has just graduated from college and plans to begin medical school in the Fall.

For National Porphyria Awareness Week, Megan recorded a brief radio announcement describing her symptoms with AIP, and informing patients that there is hope for treatment. We're so proud of Megan and her ability to deal with this disease, and we wish her every success in the future.

Good Day Houston!

APF Founder and Director Desiree Lyon and Scientific Advisory Board Chairman Dr. Karl Anderson appeared on the morning television program *Good Day Houston!* in March to discuss porphyria and the importance of diagnosing and treating the disease correctly.

Dr. Anderson runs the Porphyria Center and Laboratory at the University of Texas Medical Branch in Galveston. He has helped patients and doctors all over the world with diagnosis and treatment of all types of porphyria and runs the premiere porphyria diagnostic lab in the U.S.



Grand Rounds in Indiana

Charmaine du Toit, a nurse and APF member in Indiana, gave a seminar on porphyria in April at the hospital where she works. Charmaine illustrated her talk with a PowerPoint slide show combining the APF slides with some of her own. Charmaine is originally from South Africa, a country where Variegate Porphyria is relatively well known as the most common genetic disease.

In addition to the slide set, Charmaine made up posters, and brought the 2005 *Annals of Internal Medicine* journal article by acute porphyria experts, and the APF pamphlets.

Charmaine plans to exhibit again at the hospital's Poster Day to make doctors and nurses more aware of porphyria and the resources available to treat it.



Discovery Health: The Mystery Diagnosis Is ... EPP!

The Discovery Health network's *Mystery Diagnosis* program "The Boy Who Kept Swelling" featured a half-hour segment on Erythropoietic Protoporphyrin (EPP), including interviews with porphyria expert and APF advisory board member Dr. Maureen B. Poh-Fitzpatrick and the Leppert family. Two of the Lepperts' three children have EPP — their son, Craig, was first diagnosed at age five after several years of unexplained suffering.

EPP is a relatively rare form of porphyria, but it is the most common type found in children. The broadcast came just in time for spring, reminding all of us to cover up, and reminding physicians that if a child has classic EPP skin symptoms (painful itching and burning, sometimes accompanied by swelling and redness following sun exposure) it may be time to consider the diagnosis. The APF website has more information about EPP, its diagnosis and treatment, and preventing symptoms.

Michelle Glendenning: A Diagnosis At Last!

Michelle Glendenning has reason to be upset at the difficulty she had being diagnosed with Acute Intermittent Porphyrria (AIP). But talking with Michelle now that her condition is improving under treatment, she sounds calm and focused on doing what she can to improve her health.

Even before her hematologist sent her labs to the Porphyrria Center at University of Texas Medical Branch-Galveston for testing, Dr Anderson had already consulted in the diagnosis and care of Michelle's cousin. But Michelle found out what was making her sick only one and a half years ago — five and a half years after her first acute attack.

After being quite ill for more than five years, at age 41 Michelle finds herself debilitated. In the six months leading up to her diagnosis, she had a hysterectomy and was then hospitalized with severe attacks for five to eight days out of every month. She eventually lost the job she had held for 21 years and had to file for disability benefits.

Throughout all of this, Michelle's husband has been "amazing." When she's sick, everything stops for Michelle because she is too tired and too ill to do laundry, feed the kids or pick her youngest up from school. Her husband does double duty during these times — taking care of Michelle and visiting her in the hospital, while caring for their three children and keeping up his own work schedule.

Michelle's two stepsons are in their late teens and able to understand her condition reasonably well. But Michelle's daughter is just 10 years old, and was only seven or eight when Michelle was having repeated acute attacks and being hospitalized every month.

Explaining a serious illness to younger children is often difficult, and Michelle has the reaction any parent would: "I don't want her to have all these fears. I just want her to have a happy little childhood." Michelle points out that her ongoing acute porphyria symptoms are hard on everyone: "It's really challenging, and we just have to talk about it over and over again," to deal with her daughter's fear.

Michelle is now receiving preventive treatment with Panhematin® and improving bit by bit. While she still has attacks, she is definitely feeling better than she was two years ago.

Panhematin Update

Ovation Pharmaceuticals, Inc., the maker of Panhematin® for acute porphyria, was acquired by Lundbeck, Inc., a Danish company with international holdings, in March. Lundbeck has assured the APF that Panhematin® will remain on the market and available to patients as ever, and all physician, hospital and pharmacy access phone numbers will remain the same.



Emergency Room Guidelines: Diagnosis and Crisis Management of Acute Porphyrria

New guidelines will soon be available for emergency room doctors faced with treating acute porphyria patients. Seeking care through an ER can be particularly frightening for porphyria patients because the disease is rare and often misunderstood. It is understandable that emergency physicians, who treat thousands of heart attacks, broken bones and major traumas each year, are not always well versed in the basic checklist for treating an acute episode of a rare disease. As Desiree always says, "I don't know 6,000 people's names! How could I possibly expect a doctor to know 6,000 rare diseases?!"

Help is finally here. With support from the APF, porphyria experts are nearing completion of an easy-to-use set of guidelines for emergency room use. The guidelines spell out the hallmark symptoms of an acute attack; first-line diagnostic tests for the disease; and the immediate treatment steps necessary to prevent the patient's condition from worsening, arrest the attack and protect the patient's health.

These guidelines are different from the *Emergency Room and Primary Care Physician Kit* APF members have become familiar with. (see page 7) While the ER Kit is intended as a resource for both patients and primary care physicians, the new guidelines are for ER physicians and staff exclusively. The APF is working to distribute these guidelines to every emergency room. This will help emergency physicians administer life-saving care to porphyria patients.

Financial Aid Is Available

Getting the right treatment for acute porphyria can be difficult and expensive. If you have trouble paying for treatment with Panhematin, or for regular prescription costs and specialist doctor visits for acute porphyria, HealthWell Foundation may be able to help.



HEALTHWELL
FOUNDATION®

From financial assistance for prescription drug and doctor visit co-payments, to help with health insurance premiums, HealthWell grants funds based on medical and financial need.

HealthWell has a fund ear-marked for treatment of the acute porphyrias and Panhematin is among the drugs covered. You can apply for assistance by phone or online, and HealthWell will respond to your application within seven business days. Let's not allow money to be a barrier to your good health. Call 800-675-8416 or visit www.healthwellfoundation.org.

Protecting Your Future: A New Generation of Porphyria Experts

Dr. Brendan McGuire



The most important threat facing porphyria patients is this: we have very few porphyria experts and we MUST train more young physicians in these diseases. But training requires funding. This is why the APF established the *Protect the Future* program to train the next generation of porphyria experts. Dr. Brendan McGuire is an excellent example of the good that can come of this program.

APF members not familiar with Dr. McGuire's name will want to take note. He is a hepatologist at the University of Alabama-Birmingham working closely with Dr. Joseph Bloomer, a member of our Scientific Advisory Board.

Dr. Bloomer and Dr. McGuire treat EPP patients who have undergone liver transplant at UAB. Dr. McGuire also manages acute porphyria patients admitted to the UAB hepatology service for acute attacks, and he will be a co-investigator with Dr. Bloomer on clinical research submitted to the NIH Office of Rare Diseases Research for funding.

With Dr. Bloomer as his mentor, Dr. McGuire has the opportunity to study porphyria intensively and to produce significant contributions to scientific knowledge of and care for patients with this disease.

Dr. McGuire wrote the 2005 manuscript "Liver transplantation for erythropoietic protoporphyria liver disease," published in the journal *Liver Transplantation* in 2005, and considered to be the definitive publication on this subject. In 2008 he worked with Dr. Bloomer on the Continuing Medical Education (CME) course "The Diagnosis and Management of Acute Porphyrias," a course selected by nearly one thousand doctors as part of their required post-graduate education.

Dr. McGuire consults both nationally and internationally with physicians in need of advice on porphyria treatment and diagnosis.

Dr. Charles Lourenço

Dr. Charles Lourenço, a *Protect the Future* trainee in Brazil, will attend the *Porphyryns & Porphyrias* meeting this month in Stockholm, Sweden, together with six other *Protect the Future* doctors.

Dr. Lourenço has trained with APF advisory board member Dr. Karl Anderson at the UTMB-Galveston Porphyria Center and diagnostic laboratory. He also attended an APF educational meeting for *Protect the Future* trainees with 15 porphyria experts from the U.S. and Europe.

Dr. Lourenço has patients of his own at the Hospital Das Clinicas de Ribeirão Preto in the state of São Paulo, Brazil. With the knowledge of porphyria diagnosis and treatment he has gained through a rigorous training schedule, Dr. Lourenço also consults in the care of porphyria patients throughout Latin America, playing a life-saving role for some. The meeting in Stockholm will be an excellent next step for Dr. Lourenço's study of porphyria.



The APF is sending Dr. Lourenço to the meeting together with *Protect the Future* trainees Bradley Freilich, MD (Kansas City, MO), Chul Lee, PhD (UTMB-Galveston, TX), Lawrence Liu, MD (Mount Sinai School of Medicine, New York, NY), Gagan Sood, MD (UTMB-Galveston, TX), Manish Thapar, MD (University of Missouri, Columbia), and Jeffrey Wickliffe, PhD (Tulane University, New Orleans, LA).

We are delighted to hear that Dr. Lourenço will make a presentation to the conference this year. The *Porphyryns & Porphyrias* meeting is a scientific gathering held every two years; it is an extremely important meeting for established and new experts in the field. Physicians and researchers meet there to update each other on their latest findings on all of the porphyrias. Many APF advisory board members attend the meeting regularly as well.

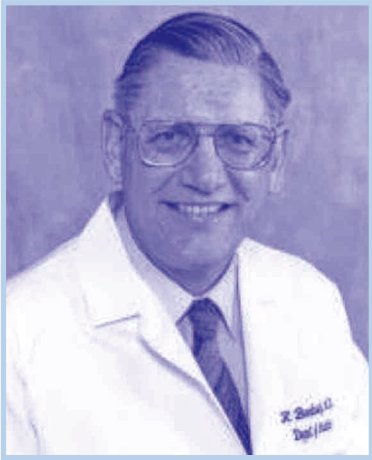
Help us Protect YOUR Future

The *Protect the Future* program insures that the knowledge of renowned porphyria specialists like Dr. Bloomer and Dr. Anderson will not be lost with their retirement. To this end, it is vital that physicians like Dr. McGuire and Dr. Lourenço have the opportunity to work side-by-side with their mentors in the study of porphyria. The training process involves an extensive course of study, lab work, patient care, and producing peer-reviewed publications.

We need a strong corps of scientists and physicians to realize our hopes for better treatment and eventually a cure for porphyria. *Protect the Future* participation, supported with grants from the APF and other sources, is vital to this mission. We are proud of the progress Dr. McGuire and Dr. Lourenço have made and we thank you, our members, for your continuing financial support. Your generosity makes this training program a reality.

To make a tax-deductible donation to the *Protect the Future* program, please call our office at 866-APF-3635 or give directly via our website, and indicate that you would like your money to go to *Protect the Future*. **Thank you.**

The Acute Porphyrias: A Lesson with Dr. Herbert Bonkovsky



On March 28, 70 APF members from every region of the country joined us by telephone for a comprehensive presentation on acute porphyria followed by a lengthy Question & Answer session with Dr. Herbert Bonkovsky.

Dr. Bonkovsky, of the Carolinas Medical Center in Charlotte, NC, has been studying and treating the porphyrias for more than 30 years and has made significant contributions

in the field. He participated in the early research on hematin, co-developed the method to prepare hematin in albumin to reduce the medication's side effects, conducted acute porphyria drug safety studies for many years and ran one of two U.S. study centers for the experimental gene therapy Porphozym several years ago. Dr. Bonkovsky is immensely popular with his patients and has served generously on the APF Scientific Advisory Board since its inception.

Dr. Bonkovsky started his presentation with a basic discussion of acute porphyria. These four disorders — ALA-Deficiency Porphyria (ADP), Acute Intermittent Porphyria (AIP), Hereditary Coproporphyrinuria (HCP) and Variegate Porphyria (VP) — are all genetic. AIP is the most common of these in the United States.

Abdominal pain is by far the most common initial symptom of an acute porphyria attack, along with nausea, vomiting and constipation. AIP and ADP are the only two types of porphyria that do not cause any skin problems, but HCP and VP can both cause a rash similar to that seen in Porphyria Cutanea Tarda (PCT). HCP, VP and PCT all cause blisters on sun-exposed areas of the skin — most often on the backs of patients' hands and arms — but they are treated very differently. HCP and VP are treated in the same way as AIP (plus protecting skin from the sun). PCT does not cause any acute attack symptoms and is the only porphyria that can be treated with phlebotomy or chloroquine.

Because so many people have questions about diagnosis, Dr. Bonkovsky discussed this topic at length. The key to diagnosing acute porphyria is measuring porphobilinogen (PBG) in urine during an attack. This is *not* the same as measuring porphyrins in blood or urine.

When a test for urinary PBG yields a diagnosis of acute porphyria, the cornerstones of managing and treating all four acute porphyrias are the same:

- 1) do no further harm — avoid drugs that can aggravate the attack;
- 2) insure adequate caloric and carbohydrate intake; and
- 3) give IV hematin to arrest the porphyria attack.

The only form of hematin available in the U.S. is Panhematin®, and it is very important to administer the medication early in an attack. Panhematin® can cause side effects, like irritation to the veins and clotting, but using a PICC line or other central catheter,

or dissolving Panhematin® in albumin rather than in sterile water, can reduce the risk of these side effects. Dr. Bonkovsky cautions that doctors should not give Panhematin® unless they are certain the patient has acute porphyria.

Once the patient has tested positive for a high urinary PBG and is being treated, further testing can determine which acute porphyria she or he has. Urine, stool and plasma tests should be performed by a laboratory with experience testing for porphyria, and the results interpreted by a porphyria specialist.

Dr. Bonkovsky discussed the fact that many other diseases can have symptoms or lab results that might be confused with acute porphyria. Many of these other conditions are also quite serious, and demand medical attention. So it is very important to receive an accurate diagnosis and administer the right care for the patient's condition.

A big danger with acute porphyria attacks is that neuro-muscular problems can worsen into muscle weakness, causing patients to lose strength in their extremities, or in the worst cases lose the ability to breathe on their own. Respiratory paralysis is life-threatening and calls for immediate hospitalization in an intensive care unit. Seizures, which a few acute porphyria patients develop, are especially dangerous when a patient has not been diagnosed because many anti-seizure medications will exacerbate the underlying porphyria attack.

Fasting can also trigger or exacerbate an acute porphyria attack, so anyone having an attack that makes it impossible for them to eat needs to be taken to the ER and admitted to a hospital, where they can be given adequate calories and carbohydrates by IV or feeding tube.

Please call the APF office or visit our website for more information on diet and nutrition, and on testing and treatment for acute porphyria. We have information for both patients and physicians, and all medical information we distribute is written by the experts of our Scientific Advisory Board. We can also help our members by putting their physicians in contact with porphyria specialists for consultation.

Doctors can visit apfdrugdatabase.com to search for drugs that may be safe or unsafe for acute porphyria patients.

*Thank You
Desiree and Dr. Bonkovsky!*

Members responded so positively to Dr. Bonkovsky's presentation that we wanted to share at least one of the thank yous we received with you:

Thanks so much for all your hard work. The phone call with Dr. Bonkovsky was such a great success for all. My husband listened and got so much from it. It's always been a worry to me that he didn't truly understand porphyria. With the conference call, he really got it! I can't thank you and all your hard working staff enough. Many blessings to all.

Scott Hall: An Early Diagnosis

Scott Hall was diagnosed with erythropoietic protoporphyria (EPP) in 1976, at age 2. His first bad reaction to the sun made his skin turn bright red with terrible itching and pain after a day spent out on a boat and picnicking in the sun. The next morning, Scott's face looked like a pumpkin and his throat was quite swollen. His pediatrician referred Scott's mother to the head of Dermatology at the University of Washington, where he was quickly diagnosed.

Scott was lucky to be diagnosed at such a young age, and his parents were lucky to find a doctor in Vancouver, BC, Canada (near their home in Washington State), who was familiar with EPP.

In addition to good medical care, his two uncles' long histories with EPP contributed to Scott's early diagnosis. Scott's uncles Wendell and Gay worked out of doors picking berries when they were young, and both men suffered for years with undiagnosed EPP. Much less was known about porphyria then and doctors speculated that the two men reacted badly to the sun because they were missing a layer of skin or had some disorder that kept them from producing enough melanin.

Of course neither of these hypotheses was correct. EPP causes sun sensitivity when protoporphyrin circulating in blood just under the skin reacts with the sun. The problem with diagnosing EPP is that while some patients' skin turns bright red and swells dramatically, others have no visible symptoms at all, only intense burning and itching.

By the time Scott was diagnosed, EPP not only had a name, but geneticists were able to find his family's specific mutation early on in his history with the disease.

Scott was extremely sun sensitive as a child but is less so now. He has taken Lumitene during the spring and summer months for most of his life to help make him less photosensitive. Living in Seattle helps too, as the weather there is overcast or rainy for much of the year.

Scott has no problems with Lumitene, although his skin does become slightly yellowed when he is taking it. He took Lumitene (formerly Solatene) even as a child, but because the pills were difficult for him to swallow, his mother had to become expert at disguising the medication. Scott reports that hiding the Solatene from him in crunchy peanut butter worked best.

Scott protects his skin from the sun and has his liver checked every year, as there is some danger of liver damage in EPP (see our website for more information).

Thanks to the efforts of APF member David McRae, Washington State law HB1000-2007 entitles Scott to a disabled parking permit because he is "restricted by a form of porphyria to the extent that the applicant would significantly benefit from a decrease in exposure to light."

If you need a permit, bring the application form at www.dol.wa.gov/vehicleregistration/parking.html to your next doctor visit, and be sure to include the physician's medical license number. Then mail or hand-deliver a copy of the form (fax copies are not accepted) to the vehicle registration office.

UCSF Honors Dr. John Epstein

The APF is proud to announce that the University of California-San Francisco Medical School is planning to raise \$2.5 million to endow a professorship in honor of Dr. John Epstein, a founding member of the APF Scientific Advisory Board.

Endowing a teaching position in his name is the medical school's way of recognizing Dr. Epstein's career-long commitment to educating young doctors.



For new MDs leaving medical school, taking everything they have learned in the classroom and translating it to symptoms they see in their patients can be a challenge. Keeping the 6,000 known rare diseases in the diagnostic picture can make this task even harder, but this is just what Dr. Epstein has done in his own work and his nearly 30 years of service as an advisor to the APF.

Dr. Epstein has authored more than 270 medical articles in his 50 years at UCSF. He has received honors from national medical societies for teaching, research and patient care. A Clinical Professor of Dermatology, Dr. Epstein specializes in photo-medicine, particularly photocarcinogenesis (sun-induced cancer), porphyria and other conditions such as allergies to the sun and lupus. He maintains a private dermatology practice in San Francisco and acts as a consultant to many of the city's hospitals.



Summer's here! Time to protect your skin

For those who get cutaneous porphyria symptoms, it's time to swing into action. Stock up on and use a good sunblock, and get covered up with protective clothing. If you do get sores or blisters, be sure to have them treated so you don't develop an infection. Let's make this a healthy season for all of us!

Member Scott Hall (see article) wears the *Sun Precautions* hat to help protect his face, sticks to long-sleeved shirts and long pants all year round and wears gloves when driving. Scott was introduced to the SPF-protective clothing maker *Sun Precautions* when he worked for the company. That time holds happy memories for Scott, since he met his wife of eight years on the job.

Contact the APF office or check our website if you need more information on protecting your skin.

The **EPP Working Kit** has information on clothing and cosmetics to protect your skin, plus expert information about EPP. The Kit is designed to help you and your physician manage your care as it relates to EPP. The Kit costs \$30 and you can order a copy by phone or on our website.

Live from Colombia: You Can Live with Porphyria!

Our dear friends and Global Partners at the Fundación Colombiana Para la Porfiria/ Colombian Porphyria Foundation are working to raise awareness of porphyria in their country, where diagnosis with the disease "can still be a death sentence," according to one recent caller from the South American country.



On May 4, Foundation members appeared in a segment called "You Can Live With Porphyria!" on the Colombian Channel RCN TV program *Cura Para el Alma* (Cure for the Soul), which deals with medical issues and spirituality.

The most important message for people with porphyria is that with an accurate diagnosis, reliable medical information, the right medications (in some cases) and good care, it is possible to treat and live with every type of porphyria. The grave danger occurs when people are undiagnosed or misdiagnosed, or when treatment is unavailable.

Porphyria Awareness programs save lives by replacing fear and uncertainty with accurate information and hope. The majority of people who inherit a gene for one of the porphyrias will never develop symptoms of the disease. The majority who do develop symptoms will go on to live normal lives. So remember: *You can live with Porphyria!*

Member Education From the APF

Dr. Bonkovsky's Acute Porphyrias conference call (see page 4) was so popular that we plan to arrange more calls like it in the future, so don't feel badly if you missed out!

In the past, the APF has invited porphyria experts to call in and speak with members at locally-organized meetings. But our members are spread out across the country and many are too geographically isolated to make attending or planning a local meeting practical. Because the conference calls let us bring many members together for invaluable education about porphyria, we are re-focusing our energies on regional or national phone calls.

We have calls coming up with Dr. Micheline Mathews-Roth and Dr. Joseph Bloomer for EPP, with a specialist To Be Announced for PCT, and with Dr. Manish Thapar for Acute Porphyria. Conference calls will give more people a chance to participate than have been able to attend the *In Touch* meetings.

We all know living with porphyria can be lonely, and this is why the *In Touch* network has always been a highly sought-after program. The APF will keep up the *In Touch* network, introducing you to others who share your diagnosis or live nearby.

Call the office toll-free at 866-APF-3635 or visit our website at www.porphyrifoundation.com for details.

In Memory

We are saddened by 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 are grateful for their thoughtfulness and desire to help others with the disease. Please join us in thanking:

La Reta Jean Roder, Boyd and Milana J. Carlson, Janyce and Gerald Kamrath, Natividad M. Horton, Carol and Kenneth Thompson, Barbara Ann Menear, Bonnie J. Clark, Frieda B. Diehl, Elizabeth and Gerald Benedict, Virginia W. Johnson, Kathryn and John Rolfe, Sandra G. Prochazka, Edwin J. Shipley, Deann and Thomas Townsend, Ruth M. Meade, Thelma and Marvin Knapp, Andy and Danny Limban, Tina Conley, Virginia Salgat, Hope and Keith Haley, Cindy H. Burton, Annette and Steven Murphy, Betty and Glenn McDaniel for **Eugene Nielsen**

Anita Miller for **Donna Pagano** and **Millie O'Toole**

Karen A. Eubanks for **Lori Brown**

C. Edwin Pearson for **Hilda Pearson**

Victoria and Matthew Gehm for **John and Judy Coley**

Diane L. Levere for **Dr. Richard D. Levere**

Gloria R. Sheehan for **Paul Sheehan**

Donald L. Johnson for **Peggy Lewis Johnson**

Dolores M. Brazas for **Wesley J. Brazas**

Janice Erickson for **Clifford and Margaret Erickson**

Linda and Rodger Fulmer for **Betty Wasson**

Janet K. Gilles for **Mildred Peters**

In Honor

Others have chosen to honor their friends through a gift to the APF:

Jennifer R. Ewing, Bette Jean and Jeff Lawrence for **Desiree Lyon Howe**

K.L. Hanson, Eric S. Gray, Gary E. Eyster for **Ralph Gray**

Rebecca L. Ross for **Betty Ross**

Debbie Smith for **Kirsten Crook**

Kathryn M. Muldowney for **Jon Pultz**

*If you wish to send a gift in honor or in memory of someone, please remember to tell us your own name and address so that we may acknowledge your gift. Please also include the name and address of the individual in whose name you are giving for **In Honor** gifts, or the name and address of the deceased's loved one for **In Memory** gifts, so that we may inform them of your kindness and sympathy. Thank you.*

Our deepest sympathies

The APF extends its condolences to member *Lisa Kancsar* and her family, who mourn the loss of Lisa's father. *Eugene Nielsen* died in February following a battle with double pneumonia and Alzheimer's disease. Although Mr. Nielsen did not have AIP, Lisa writes "I know he would have desired for the APF to benefit from any donations in lieu of flowers."

On behalf of all our members, we thank you Lisa. We promise to honor your father's memory by enhancing public awareness about this disease, educating patients and their doctors, and continuing to support physician training and research for a cure.

Dr. Cecil Watson: Father of Porphyrins and Porphyrins in the U.S.



I am grateful to Dr. Sylvia Bottomley for what I've learned about porphyria history since her gentle correction to our last newsletter.

The Spring issue of the APF newsletter referred to Dr. Cecil Watson merely as a developer of the Watson-Schwartz screening test for the acute porphyrias. Yet as Dr. Bottomley pointed out, Dr. Watson is in fact rightly remembered as a luminary in American academic medicine and the father of porphyrins and porphyria studies in the United States.

Dr. Watson graduated from medical school in 1925, and from 1930-32 studied tetrapyrrole chemistry under the Nobel Prize winner Hans Fischer in Germany. Dr. Watson went on to chair the Department of Medicine at the University of Minnesota while it grew into one of the major medical schools in the U.S. He played an important part in the developing National Institutes of Health as well.

In an appreciation of Dr. Watson's work published in 2005, Dr. Rudi Schmid, a giant among porphyria researchers himself, notes that it was in Watson's lab where he became the first to succeed in "chemically inducing full-fledged hepatic porphyria in experimental animals" — a vital step in scientific understanding of how these diseases work. And Dr. Bottomley notes, "many of us working in porphyria today knew this legendary man in person."

Among Dr. Watson's 350 medical publications were major articles on the use of hematin infusions to repress the biochemical overproduction cycle at the root of acute porphyria and treat acute attacks. Dr. Watson died in 1983, shortly after Panhematin, the life-saving medication whose development he had pioneered, was approved by the FDA.

The APF and our members owe an enormous debt to Dr. Watson — both for his own scientific work on all the porphyrias, and for seeing so many of the experts we rely on today on the road to understanding these rare and complicated diseases.

— **Mira Geffner**

Correction: Dr. Sylvia Bottomley

Dr. Sylvia Bottomley, whose most recent revised chapter on the porphyrias was published in *Wintrobe's Clinical Hematology (12th Ed., 2008)* last December, is Professor Emeritus of Medicine at the University of Oklahoma Health Sciences Center in Oklahoma City. We omitted this information from our article about Dr. Bottomley in our last newsletter. Dr. Bottomley and other porphyria experts are available for consultation with physicians seeking advice on diagnosing or treating porphyria. *Members can contact the APF office or ask your doctor to call us for details on consulting an expert.*

If it looks too good to be true . . .

Finding a diagnosis can be difficult when you have a rare disease, and it can be challenging to find reliable testing for porphyria. When you put these things together, patients who suspect they have porphyria can run into some pretty big problems. Sometimes even with a clear diagnosis it can be hard to get the right treatment — if your doctor or insurance company is not familiar with porphyria, for example, or if the doctor's office or hospital is very far from home or work.

Unfortunately, there are some websites offering unproven tests and treatments for porphyria directly to consumers. These products are available on the internet and may not be backed by expert medical or scientific research. This means they might not be of much help to patients, and they could potentially do more harm than good.

The APF advisory board of porphyria experts recommends that physicians ordering tests for porphyria rely on a laboratory with experience in diagnosing the porphyrias — preferably one overseen by an expert who can offer reliable interpretation of the test results. The APF website has detailed information on how and where to test for porphyria and on treatments for each of the eight porphyrias.

If you have trouble finding information about porphyria, just call our office — we're here to help! We also offer our members' doctors a *Physician Education Kit* with treatment and testing information from the experts, free of charge.

Remember: it is always a good idea to discuss any treatment or testing with your physician. And if it looks too good to be true, it probably is.

What You Need in an Emergency

For all people with acute porphyria, preventing attacks is crucial to insuring the best possible long-term health. Luckily, the APF has something that can help: the **Primary Care Physician and Emergency Room Kit**.

The kit contains information to help your regular doctor keep you healthy and basic information to guide an ER doctor managing your care in a crisis:

- *Annals of Internal Medicine* review article on acute porphyria;
- A step-by-step guide to diagnosing porphyria, lab instructions and a list of U.S. porphyria laboratories;
- APF pamphlets on Panhematin treatment and managing acute porphyria;
- Instructions for preparing Panhematin for infusion;
- Rapid PBG testing kit information (kit for laboratory use only);
- A list of safe/unsafe drugs for use in the acute porphyrias.

The kit also has a place for your personal medical records.

The ER/Primary Care Physician Kit is available from the APF for \$30. All APF brochures are also available for individual purchase, and all medical information we distribute is written by porphyria specialists.





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

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

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

Updated Member Stories Section: Full-length versions of the member stories in our newsletter.

A free one-credit online **Continuing Medical Education** course on the acute porphyrias for physicians.

Tell your doctor about the **Safe/Unsafe Drug Database for Acute Porphyria** at www.apfdrugdatabase.com/ and let him or her know all the medical information we distribute is written by long-time porphyria specialists.

Is Your Membership Up to Date? Stay current on the latest news about testing, treatment, and upcoming events. Please take a moment to renew at our website, or call us at the office: 713-266-9617 or 866-APF-3635. *Thank you.*

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