



Dear Friends, Yvette, Elizabeth, Lelia, Carol and Desiree wish you and yours a very Merry Christmas and Happy Holiday season. We pray that your lives will be filled with happiness and good health throughout the New Year. We also want to thank you for the many kind and encouraging phone calls and letters we have received. Meeting so many of you has been a wonderful blessing in our lives.

Together we have contributed to the well-being of thousands of people who rely on the APF for education about porphyria, to support research, to enhance awareness within the medical community and to receive help in their greatest time of need. We look forward to the coming New Year and the many opportunities we will share to achieve a happy, healthy future and ultimately a cure for

porphyria. God Bless each of you with His love, joy and peace.

Interested In Meeting Other APF Members? Attend or Host An In Touch Meeting

One of the ways we share our lives and work together toward a healthy future is through your participation in our IN TOUCH network, which is a program where we can communicate with each other and/or meet one another at regional gatherings. If you are interested hosting an APF IN TOUCH Meeting in your area, we are here to help. Your meetings can be a simple night or weekend gathering in your own home. People with porphyria are anxious to communicate with one another and learn from each other, so they enjoy almost any venue that allows them to find such camaraderie. Many churches, libraries, etc. will allow you to hold a meeting for free if it is not convenient to host a meeting in your home. The important element to have a successful meeting is to provide a relaxed, setting where people can enjoy themselves, and the APF will help facilitate the meeting and provide the members and the materials to make it a success. In fact, we often orchestrate a special conference call with one of the porphyria specialists during your meeting. We also will send you the "Porphyria Live" DVD to use as a focus for your meeting, which everyone seems to enjoy.

Dave McRae, host of the Seattle meeting, describes it, "The meeting, although small, was really amazing. It is so nice to be able to talk with other porphyria people. We introduced ourselves and told each other about life with this disease. The stories were both heartwarming and heartbreaking. We talked like we had known each other our whole lives. We ended the meeting by watching the DVD which we all enjoyed. My wife, Laurie Prendez, did most of the work and was very involved in the conversations. We would like to meet again in a few months."

April Henderson, who hosted the Northern California group, said, "Wow, it was perfect! We made all made an awesome connection! I'd planned to start the meeting with the DVD, but instead we began by introducing ourselves and talking a little about what we hoped to achieve by meeting together. It was very touching, and I think it really helped us to relax and bond. Then we took a few minutes to list the topics we wanted most to address with the Dr. Claus Pierach, a respected porphyria expert. Hunter from Ovation Pharmaceuticals facilitated the conference call with Dr. Pierach, who was truly delightful, charming, helpful, funny, warm, courteous, and patient. He not only welcomed, but even encouraged us to ASK, ASK, ASK! Everyone thanked Dr. Pierach and Hunter for making time to attend, and we chatted a bit after the call to help absorb what we had just heard. Next, we turned on the DVD. It was a HUGE hit, too! We came away from the meeting feeling more empowered and happy to have made new friends in the porphyria community. Plus, right then and there we all readily agreed to keep "in touch", and we are already planning our 2nd meeting!" On behalf of our group, I want to thank Lelia and everyone at APF for all their hard work, information and support. Such comments are frequent after the meetings by both hosts and guests. If you would like to join the IN Touch Network, please contact the APF office 713-266-9617 or elizabethapf@aol.com, and if you would like to host a meeting, please contact Lelia Brougher, our IN TOUCH coordinator, at email@broughers.com.



Dr. Joseph Bloomer

Another famous name in the world of porphyria is Dr. Joseph Bloomer. Dr Bloomer is the Director of the Liver Center Section and head of the Hepatology Department at the University of Alabama in Birmingham. Dr. Bloomer received his undergraduate degree from the Massachusetts Institute of Technology and his

M.D. degree from Western Reserve Medical School. Following a medical internship and residency at the University of California Hospital in San Francisco, he spent three years at the National Institutes of Health as a Clinical Associate, where he developed his interest in liver diseases. Dr. Bloomer next attended Yale University School of Medicine as a postdoctoral fellow in liver diseases and later joined the Yale Liver Unit as a faculty member and was then appointed Professor of Medicine and Director of Hepatology at the University of Minnesota. There he also directed a Digestive Diseases Center which focused on advanced liver disease and porphyria. In 1995, he was recruited to the University of Alabama at Birmingham as Director of the Liver Center and Section Head of Hepatology. Dr. Bloomer is a former Howard Hughes Investigator and a member of several scientific and professional organizations. He is currently supported by a MERIT award from the National Institutes of Health and recently served as President of the American Association for the Study of Liver Diseases (1998-1999).

Dr. Bloomer has received NIH research support since 1976 to investigate the pathogenesis of clinical and biochemical features in the porphyrias. His laboratory was the first to define the enzyme defects in two of these eight human genetic disorders, erythropoietic protoporphyria (EPP) and variegate porphyria, and the first to show that liver disease in EPP is caused by the toxic effect of protoporphyrin on liver structure and function.

Most recently his laboratory has demonstrated that symptomatic disease in EPP is associated with a mutation in one ferrochelatase allele. This has profound implications regarding genetic counseling that is given to families with EPP and for gene therapy which may be developed for patients with severe disease. Molecular features of EPP liver disease in humans are also being examined by DNA

microarray analysis of hepatic genes in liver explants of patients who have undergone transplantation.

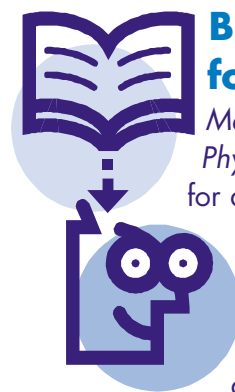
Dr Bloomer also participated in writing the *Annals of Internal Medicine* article and was the primary author of the new Continuing Medical Education (CME) course for physicians for the management of the acute porphyrias.

A Family Affair

Members of the Pagano/O'Toole family have been members and supporters of the APF for many years. They have contributed in various ways but one way in particular is by honoring their family and friends with gift cards through the IN HONOR and IN MEMORY gift program of the APF. When someone in the family has a birthday or is deceased, some of the family members have sent a gift in their honor. In turn, the APF notifies the honoree or the family of the deceased that their family or friend has been remembered with a gift of the APF cards.

They also try to support and educate one another since porphyria runs in their families. At a recent family reunion, they showed the *Porphyria Live* APF DVD. The film was a big success. What a terrific idea, particularly since porphyria is inherited.

Other members have shared the DVD at IN TOUCH meetings and medical seminars. The importance of enhancing awareness of the disease in your own family cannot be stressed enough and the DVD is an excellent tool to use. You can purchase a copy of this outstanding DVD for \$20.00



Brochures Available for Members

Management of the Acute Porphyrias, A Physician's Guide is the new APF brochure for diagnosing and treating each of the acute porphyrias: AIP, HCP, VP and ALAD. The publication of this brochure is part of our expanded Physician and Patient Education program. These brochures are being distributed to the physicians on our database. If you have not given us the name and address of your physician, please do so as soon as possible. We will then forward not only the new brochure but also a copy of the *Annals of Internal Medicine* article which describes the diagnosis and treatment options in more detail. You can order several for yourself and your other physicians for \$1.30 each.

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L-R: Anne, Linda, John, Desiree and Dick planting trees as a fundraising event. We would like to have photos of your events.

NATIONAL PORPHYRIA AWARENESS WEEK

National Porphyria Awareness Week is February 18 – 25, 2006. Although February seems far in the distance, it will be here soon. We need to be prepared for this big event if we don't want it to slip quietly by unnoticed. You can play a major part to enhance awareness of porphyria within your own community and around the country through the many services and programs of the American Porphyria Foundation. **Together** we can make the **National Porphyria Awareness Week** an extraordinary event. By spreading information about porphyria, the public and physicians alike will more readily recognize the need for better diagnosis and treatment. Here are a few suggestions for your participation.

Support the APF and You Support Each Other!

Become an APF Ambassador by sharing information about porphyria wherever you can.

Refer individuals or families living with porphyria to the APF and/or its website.

Join the IN-TOUCH Network as a means to discover new friends and share knowledge.

Make a difference in porphyria awareness, education and research through your donations.

Purchase one of our outstanding educational books, brochures, DVD's.

Volunteer your talents to help enhance the APF newsletter, create new educational materials, use for fund raising projects, etc.

Sign up with your employer for a matching gift for the APF or sharing programs like United Way.

Assist at an exhibit booth at a medical convention or distribute medical information in your community.

Suggest that family and friends make a donation to the APF in honor of a birthday, anniversary, or memorial.

For more information on how to participate in the **National Porphyria Awareness Week**, please contact Desiree at Lyonapf@aol.com or call the APF 713-266-9617 or Tonya Love, the National Porphyria Awareness Week Coordinator at 205-620-3855 or TonyaAIP@aol.com.



Study Helps Explain Why Fasting Brings On Attacks

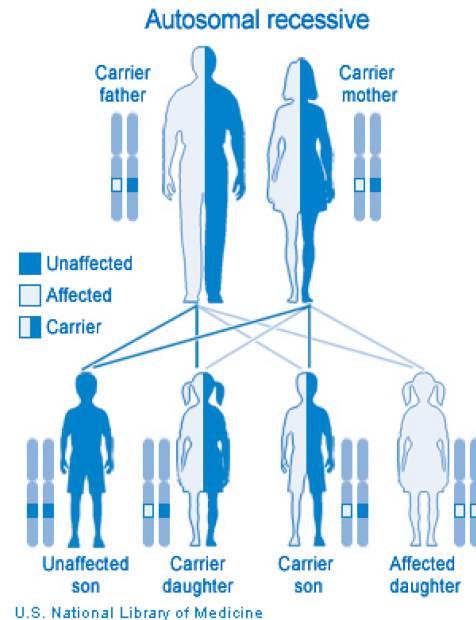
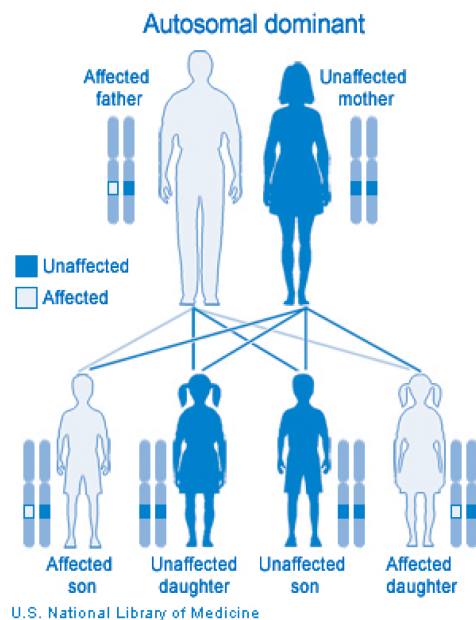
A team of researchers headed by Dr. Bruce Spiegelman of the Dana Farber Cancer Institute and Harvard Medical School has discovered a molecular missing link that helps explain why fasting brings on attacks of the acute porphyrias. The finding could help improve treatments. Researchers and physicians have long known that fasting can cause acute attacks of the disease and that the attacks can be relieved with glucose or other high-carbohydrate treatments. However, the exact link between fasting and the attacks has been mysterious until now. Spiegelman's study shows that fasting increases levels of a metabolic protein called PGC-1a. The "starvation" signal that fasting sends throughout the body prompts PGC-1a to jump-start the process of creating glucose from scratch in the liver. However, PGC-1a also regulates the activity of an enzyme called ALAS-1, the first key enzyme in the heme production pathway. The higher levels of PGC-1a produce higher levels

of ALAS-1, leading to a toxic buildup of precursor heme molecules, Spiegelman and colleagues found. The finding explains why glucose infusions are helpful in treating acute attacks since the glucose boost can shut off the starvation signal and return PGC-1a levels back to normal.

However, the discovery could pave the way for new porphyria therapies that focus on PGC-1a itself rather than relying on high-carbohydrate treatments that often cause weight gain, the researchers suggest.

Charitable Medical Air Transportation

The National Patient Air Transportation Helpline is the only service of its kind in America; specializing in helping patients find free or low cost travel and hospital hospitality for family members in distant cities far from home. It can be reached by calling 1-800-296-1217. This help line is operated by Mercy Medical in support of Angel Flight America, which does 90% of the long distance charitable medical air transportation.



When a genetic disorder is diagnosed in a family, family members often want to know the likelihood that they or their children will develop the condition. This can be difficult to predict in some cases because many factors influence a person's chances. One important factor is how the condition is inherited. For example:

- A person affected by an autosomal dominant disorder has a 50-percent chance of passing the mutated gene to each child. There is also a 50-percent chance that a child will not inherit the mutated gene (*illustration*). AIP, VP, HCP, PCT, ALAD, EPP
- For an autosomal recessive disorder, two unaffected people who each carry one copy of the mutated gene (carriers) have a 25-percent chance with each pregnancy of having a child affected by the disorder. There is a 75-percent chance with each pregnancy that a child will be unaffected (*illustration*). CEP

To continue, if a couple has a child with an autosomal recessive disorder, the chance of having another child with the disorder is still 25 percent (or 1 in 4). Having one child with a disorder does not "protect" future children from inheriting the condition. Conversely, having a child without the condition does not mean that future children will definitely be affected. Although the chances of inheriting a genetic condition appear straightforward, in some cases factors such as a person's family history and the results of genetic testing can modify those chances. In addition, some people with a disease-causing mutation never develop any health problems or may experience only mild symptoms of the disorder. If a disease that runs in a family does not have a clear-cut inheritance pattern, predicting the likelihood that a person will develop the condition can be particularly difficult. Because estimating the chance of developing or passing on a genetic disorder can be complex, genetics professionals can help people understand these chances and make informed decisions about their health.



Attacks and International Travel

According to Dr. T.J. Peters and Dr. D.J. Deacon at Department of Clinical Biochemistry, King's College Hospital, in London, five patients were reported with acute intermittent porphyria in whom attacks were apparently precipitated by international air travel. In four patients, international travel was the initial presenting attack and in a fifth person, it was the cause of an acute relapse in a patient requiring regular heme therapy prophylaxis. Multifactorial precipitants implicated include, dehydration, missed meals, alcohol use, infection, chronic hypoxia, premenstrual syndrome and stress. Thus, acute intermittent porphyria should be suspected in individuals with unexplained acute abdominal pain following international air travel. Appropriate precautions may reduce the incidence of attacks.



Melanie Robinson

Before I start my story, I want to tell everyone who is involved in the APF and its website, "THANK YOU," and if someone with porphyria is reading this, "Don't be scared! You really can live with it!"

I am 23 years old and was in perfect health when my

ordeal started, I had just graduated from college with a degree in Biology, Nursing, and Theatre. I was in my first semester of graduate school (Master's in Nursing) and just started my new job as an RN on an oncology unit. Shortly thereafter, I had a sinus infection and became too nauseated to eat. I also felt like someone kicked me in the stomach but assumed it was pre-menstrual cramps, so I ignored it. I dealt with this horrible abdominal pain for a week before I was sent home from work because I kept vomiting. I visited a doctor, who gave me a urine test for analysis. It was then that I realized how dark my urine was. The nurse told me I was dehydrated, and the doctor told me I had a stomach virus. He said that I didn't need anything for the pain, because it would mask my symptoms and if they got worse, I should go to the ER.

The pain continued, so I went to the ER. I was told that I possibly had an STD, ruptured ovarian cysts, appendicitis, or pancreatitis. After all of the tests, I was sent home after 20 hours in the ER. The doctor gave me vicodin for pain and told me that I should feel better. They insisted that the final diagnosis was an STD even though they never took a vaginal swab and even though I was on IV antibiotics for the whole 20 hours in the ER. The pain didn't even dwindle except with the first one or two hours after the morphine.

Two days later I was still crying in pain, and I went back to the ER and was admitted. The doctors found nothing. I had a colonoscopy, NG tube inserted, Foley catheter, exploratory laparoscopic surgery, and after a week, I went sent home with a referral to a psychiatrist. I was told I was too stressed out, it was in my head, or maybe I was drug dependent. I was a straight A student and a stable individual, yet the doctors all thought I was crazy!

I felt better for a month or two and then the dreadful pain returned. Once again the doctor refused to admit me and gave me Vicodin, but I kept vomiting and was finally

admitted me to the hospital again. A GYN saw me and said that I never had an STD, (which I knew already) and that maybe I should be tested for sickle cell anemia and something called Acute Intermittent Porphyria. Finally, someone believed me!

I was depressed, angry, scared but finally grateful. The APF has helped me so very much. The struggles of the people before me lessened my suffering. If it wasn't for this foundation and their attempts at increasing awareness, I probably would still be suffering in pain.

I never thought I would feel fine again, but now I am leading a normal life and am back at work and am eating what I call the "Anti-Atkins" diet. I have learned to take nothing for granted and to listen to my patients when they tell me their symptoms. Four people in my hospital have now been tested for porphyria. Hopefully, I have done my little part in increasing awareness! Thank you again!

Name Our New Radio Show



Phil Grohs, a long time APF member, has provided us with the most amazing opportunity through his **Live 365** radio program. By downloading a simple, free program on your

computer, you can listen to a lecture given by a porphyria specialist or similar medical authority or even grace the airwaves with your own story. You can be the Star of the new APF weekly radio show! Name our new radio show and win a year's APF membership, Desiree's book, an ER kit and the *Porphyria Live* DVD, a \$100 value.

You will receive or have already received directions on how you can enter the site and listen to the sessions and will receive notice about the time of the upcoming sessions on the internet. You can tune in for the speaker at the appointed time. But how will we all be able to hear your story. This is easy, too. We are asking you to submit a cassette tape in your own voice reading or telling your porphyria story. Then we will broadcast your story after advertising the event via our internet Enews. Just go to www.live365.com/stations/zipper240z?play for details and be sure to send in your suggestion for the Name of the radio show. If you have technical issues to enter the site, please contact Phil at ol_southerner@hotmail.com.

Wendy's Walk

Wendy Ver Voort has been a member of the APF for many years. Recently she completed a grueling walk for the second time to help raise awareness of porphyria, as well as funds for much needed research. Her ultimate goal is to keep others from the suffering she has endured. "I spend anywhere from three to ten times a year in the hospital. It's been a little bit rough because, I've noticed how much more nerve damage I have since the last time I did the walk about seven years ago, but if it helps someone else, it is worth it." Thanks Wendy

In Wendy's words:

I made it! The total walk was 141 miles, and the temperatures were in the 80's and 90's all week. We won't talk about what my feet looked like or how burnt I was. I would sweat off the sunscreen as soon as it went on, and by day six I couldn't get shoes on anymore. Walking 14 miles that day with bare feet wasn't my brightest idea I've ever had, but I was bound and determined to make it. The last day my sister picked me up some "Crocs" (best shoes ever made). I could have kept going another 100 miles.

I was very happy with the awareness that I received: two news stations, the local newspaper and a radio station that broadcasted about the walk several times each day. "Bear," a well known DJ, even came out and walked five miles with me one day.

About a month before the walk I had an attack. I was in the hospital for three days, but was back in six hours later for surgery. Needless to say, I was a little nervous I wouldn't be able to do the walk; but God was with me. My three boys did say though, that next time they would be doing the walking; and I'd be driving the relief vehicle. I probably



Wendy ver Voort

won't argue. I also received a nice letter from a woman who lives in Arizona and heard about it. Arizona! Can you believe it, a long ways from Wisconsin. My pledges for the APF have started coming in, so I am sending what I have with this letter along with some pictures from the walk. Sincerely, Wendy ver Voort

Medical Conventions

We are exhibiting at two medical conventions: The American Society of Hematology to be held in Atlanta and the American Association for the Study of Liver Diseases, which was held in San Francisco. Both are important disciplines for porphyria treatment and research.

Leila Brougner, convention coordinator, and APF members, Rose Jeans, Gabriella Buttz, Angelina Buttz, Geoffrey Sarphati, Danielle Frazzini Tarka, Stephani Frazzini, John Hurley, and Jennifer Predmore will be at the APF booth to distribute educational material to physicians and other medical professionals. These conventions, which are attended by approximately 10,000 doctors, are a primary means to educate doctors about porphyria. Your help is needed when they are held in your area.

Gear up for National Porphyria Awareness Week

**February 18-25, 2006 and
the 25th Birthday Celebration of the APF!**

Be the first to order your American Porphyria Foundation T-Shirt and Baseball Cap for yourself and gifts for family and friends.

The 100% white cotton T-shirt will have the APF name, logo and National Porphyria Awareness Week dates. The sporty Baseball Cap is either crimson or blue and embroidered in white with the APF name and logo. Please be sure to note the color of the cap that you would like on your order.

Respond today in preparation of 2006 National Porphyria Awareness Week and order T-Shirts and/or Ball Caps. Just call the APF office with your credit card number at 713.266.9617 or mail your order with check, cashiers check or money order.

Baseball Hats – **\$12.00 + \$3.85 shipping = \$15.85**

T-Shirts – Adult sizes S through XL

\$15.00 + \$3.85 shipping = \$18.85

T-Shirts – Adult size XXL

\$20.00 + \$3.85 shipping = \$23.85



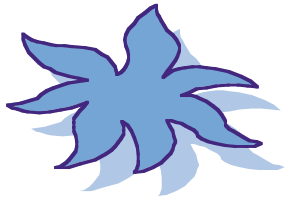
APF Partner Award

Last year, the APF established an annual award for the person or company who has greatly assisted us in accomplishing the mission of the APF—to enhance patient and physician education.

The first award was given to Mr. Jeffrey S. Aronin, President and CEO of Ovation Pharmaceuticals.

This year's APF Partner Award is

given to APF member **Tonya Love** for her exceptional commitment to aid people with porphyria. Tonya has volunteered countless hours to assist us with media projects, computer research, In Touch projects, physician education programs, and compassionate support to her fellow members. In fact, Tonya established the National Porphyria Awareness Week and the monthly internet "chat" program and will continue to oversee both programs in 2006. I know that you join me in thanking Tonya for her dedicated service.



Some Plants Can Fix Their Own Genes

Geneticists at Purdue University have found that some plants can replace their defective genes, as if

they have back-up copies from former generations that can bypass the usual mechanisms of heredity. The discovery has been described as "really strange and unexpected." It defies Mendel's laws of inheritance. Evolutionary biologists have long believed that genetic mutations have led to the evolution of different species. If plants and animals can correct their own mutations, it poses problems for evolutionary theory.

Scientists were studying a mustard-like plant called *arabidopsis* with a mutated gene that made the plants' petals clump together. The plants had two copies of the mutated gene, so they had no chance of having normal offspring. However, ten percent of the plants' offspring kept reverting to normal. When the scientists analyzed the mutated genes in the offspring, they found they had changed back to normal genes. Much more research is needed to understand why this happened and whether it could occur in other plants and animals. Understanding how genes can be changed will hopefully lead to human medical applications.

In His Memory

Sharon Woznicki and her sister, Kathleen Marsh and their family honored their father, James R. McDougall, with donations to the APF [In His Memory Fund](#). Mr. McDougall had AIP, but recently passed away after a long battle with cancer.

Their family felt that the American Porphyria Foundation could use the donations to continue research for treatments and ultimately a cure for this disorder. Mr. McDougall and Kathleen proudly participated in drug studies for heme arginate, hemein, and tin mesoporphyrin through the University of Texas at Galveston. He felt that volunteering for research was a way to help save his daughter from her many severe attacks since hers is the worst case in their family. His participation in the study was just another way for him to show how much he cared for his family. Mr. McDougall was the type of man who would do anything to help other people as well.

We sincerely want to express our sympathy to the McDougall family over their loss, and we think them for honoring James through gifts to the APF. *Thank you.*

You, too, can remember family and friends

You can honor the memory of a loved one or a friend through a gift to the APF. We will then send a memorial card in your name to the family of a deceased friend or loved one for a donation of \$10 or more. When making a memorial gift, please send us your name and address, as well as the name and address of the family to whom the card should be sent. Remember to tell us the name of the individual you wish to honor or memorialize. Gifts may also be made in honor of someone on a special occasion. The amount of the gift is not revealed unless you request to do so.

Continued (Brochures Available...) from page 2

Also available are the following other brochures that can be purchased for \$1.30 each: **[Overview](#)** explains each type of porphyria, including diagnosis and treatment; **[Panhematin](#)** describes why, when and how Panhematin is used to treat the acute porphyrias; **[Diet and Nutrition](#)** helps determine what nutritional measures are important for AIP, HCP, VP, ALAD; **[Drugs and Porphyria](#)** relates drugs and environmental agents that precipitate acute porphyrias; **[Porphyria Cutanea Tarda](#)** explains this photosensitive type of porphyria; **[Erythropoietic Protoporphyrin](#)** explains this photosensitive type of porphyria.



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 on the APF Web Site

www.porphyrifoundation.com

Be the First to order the APF brochures: The new management brochure for the acute porphyrias is available, as well as brochures for diet, drugs, pct, epp, panhematin and general information.

Be the First to order your Awareness Week T-shirts and Caps: You can order cotton Tshirts with the APF logo announcing the National Porphyria Awareness Week.

Be the First to volunteer to host a National Porphyria Awareness Week meeting: You can host a meeting for other APF members in your community or with the local medical community. You can also participate by giving an interview to your area newspaper or television station or facilitating a fund raiser.

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