



Important Notice for Your Physicians

Recommendations for the Diagnosis and Treatment of the Acute Porphyrias

is the first Continuing Medical Education (**CME**) program for physicians on the subject. This fully accredited course is now available free of charge and can be completed via the "For Physicians" section of the American Porphyria Foundation via: www.porphyrifoundation.com or www.acuteporphyras.net.

At the completion of this **CME** activity, the participants should be able to:

- Recognize the symptoms of the acute porphyrias.
- Diagnose and test for acute porphyrias.
- Evaluate therapeutic options.
- Manage patients suffering from frequent acute attacks.

The course is sponsored by Rush University Medical Center, which is accredited by the Accreditation Council for Continuing Medical Education to provide continuing medical education for physicians. Rush University Medical Center designates this educational activity for a maximum of one credit of category 1 toward the AMA Physician's Recognition Award.

If you have one of the acute porphyrias, AIP, VP, HCP or ALAD, please tell your physician about this most important opportunity. In addition, if you will provide your physicians name and address, we will send him/her more information about this **CME** course. Please contact Yvette or Elizabeth at the American Porphyria Foundation: 713-266-9617, porphyrus@aol.com.

Lyon's Share — Research Volunteers Needed

One of the most gratifying experiences of my life has been as a research volunteer. Today many of our APF members are taking treatments and medications that I and many other research volunteers were given as part of a research project. In fact, I have participated in seven research stud-

ies at NIH and Rockefeller University and have been blessed to play some part in helping other people with porphyria. I know that all of you who have had this opportunity feel the same way.

Now we are in very great need of participants to research Porphozym™, the enzyme replacement therapy, produced by the Danish company, Zymenex, formerly Heme Biothec.



Jennifer Gattoni, (third to right) Porphozym research participant, is joined by her husband, mother and mother-in-law.

It is currently in international Phase IIb clinical trials for the treatment of AIP. The researchers for this study, Dr. Karl Anderson and Dr. Herbert Bonkovsky, are not able to complete the study without you. The parameters for this new study are: the patient must have documented AIP, frequent attacks and specific biochemical test levels.

Every research project that is FDA approved, whether it be for cancer, heart or porphyria, has a very clear, consistent protocol that applies to every person who volunteers to be part of the study. Therefore, when the physician/investigators applied for approval to undertake the study of Porphozym™, they had to submit and agree to follow very strict guidelines before they were given the FDA approval. As mentioned previously, the protocol for the enzyme replacement includes: the research volunteers must have documented AIP, frequent attacks and specific bio-chemical tests levels which can not be changed. Without such strict parameters, there would be no way of determining if Porphozym™, or any other treatment is effective.

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Carbohydrates

For some individuals who have the "acute porphyrias" (AIP, VP HCP and ALAD), attacks can be brought on if carbohydrates and calories are restricted for prolonged periods of time. For example, during the Atkins Diet

craze, many people with porphyria were diagnosed when their reduction of carbohydrates precipitated attacks. This is why fasting or major dieting is not recommended. Thus, to prevent and treat attacks, carefully monitoring one's diet can be especially important for these types of porphyria.

Why are these three porphyrias more sensitive to diet? The pathway in the liver that makes heme from porphyrins and other substances is very sensitive to carbohydrates in particular. Therefore, when less carbohydrate is taken, it appears that porphyrin production is stimulated, and the body can't use them all effectively. This porphyrin overflow is what creates the symptoms of an attack.

Carbohydrates are the foods that contain starches or sugars. They are important in everyone's diet, because they provide us with fuel for our bodies, as well as a wide variety of vitamins and minerals in many of the carbohydrates.

Starches are "complex carbohydrates" and tend not to be as sweet as sugars. Starches are called "complex" because they are larger molecules and take longer (compared to sugars) for our bodies to break them down for use as an energy source. Some starchy types of foods include: potatoes, pasta and bread.

Sugars are "simple carbohydrates", meaning their molecules are not that big and are quickly broken down in the body. They are quickly absorbed into our bodies as an energy source. These sugars may be found naturally in foods, such as fruit, fruit juice, some vegetables, and milk and milk products. Sugars are also found in higher levels in foods like honey, table sugar, candy, syrups and regular soda pop. Note that this last group of foods, although high in sugar, lack vitamins, minerals and fiber. Starches and sugars eventually break down into a substance called glucose, which is used as fuel by the body.

Carbohydrates often play a paramount role in preventing and treating an acute porphyria attack. When carbohydrates are broken down into glucose in the body, it may

help minimize the over-production of porphyrins in the liver.

Because this seems to be such a simple treatment, it is not adhered to by some members who don't understand that this simple treatment involves a very complex mechanism. The guidelines suggest that when a person is having symptoms of porphyria, the carbohydrate intake be increased. In fact, some people help prevent attacks by maintaining a high carbohydrate intake daily.

The Heme Pool

Imagine that the pathway is actually a river of porphyrins and porphyrin substances which together flow into and make up a big red-coloured pool called the Heme Pool. Like a recipe, various ingredients (or porphyrin substances) are added to this "river" at various stages to form the Heme Pool. Along this river there are several gates (which represent different stages of the Heme Pathway). These gates must be opened fully by an operator in order to keep the proper flow of the river into the Heme Pool. Each gate has a different operator that has a unique key (which we'll call an Enzyme Key) for the gate that he must open. The job of each operator is to insert their Enzyme Key into their gate, and to turn the key fully so that their gate will only open part way, thus decreasing the flow of the river, resulting in a lower level in the Heme Pool.

One day the operator at the third gate comes to work and is not feeling well. Because of past incidences, we know that when he wasn't well, he had taken medications that didn't agree with him; drank alcohol; didn't eat all day; or got too much sun. One or two of these things seemed to affect how he did his job...specifically how far he could turn the Enzyme Key in the gate. When he had exposure to these "irritants" before, it could not open fully. Regardless, he still has to go to work, since no one else can operate his gate. Unfortunately, because his gate doesn't open fully, the flow of porphyrins start the Pool to tell the first gate operator to turn up the porphyrin tap to increase flow. "Heme levels are decreasing!" they say, not even aware that the third gate operator isn't doing his job properly. This keeps continuing until there is a flood at the third gate. This flood puts the whole flow into chaos until the operator at the third gate can get back to normal. For this particular gatekeeper in the past, having him take sugar often helped him get back on track. In a worst case scenario, a specialist would have to be brought in to help get the third gate operator back to

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IN TOUCH MEETINGS

We have had a number of IN TOUCH MEETINGS around the country. If you are interested in hosting a meeting in your area, contact Lelia Brougher, our IN TOUCH Coordinator at email@broughers.com or 404 550-4880. You will be notified of a meeting in your area or check the APF website for information about upcoming meetings. The following are reports from some of these meetings. If you have held a meeting, we would like to place a story about the meeting in the newsletter or on the APF website.

Colorado Meeting Five strangers met on May 18th. It didn't take but a few minutes for them to be talking, laughing and listening as though they were old friends. Sharing experiences with a common challenge somehow made that happen. Mary Gibbs, who has been dealing with her porphyria for many years said, "I thought I was alone in this. It was wonderful to meet other people with porphyria and feel their support. It is encouraging to share with others."

It was a pleasure for me to host this meeting, and it was made easy because of the help from Lelia Brougher from the APF, Wendy Schoal from the Ovation Pharmaceutical Company and Dr Claus Pierach, who has been a renown specialist for thirty years. It was indeed encouraging to hear Dr Pierach's (we had a telephone conference call with him) commitment to improving the lives of people with this disorder. Three spouses attended the meeting and were heard sharing their unique experiences as loved ones and care givers of someone with porphyria.

As we parted at the end of an interesting and rewarding evening, we all expressed our intentions to meet again soon. As of now, we are trying to find a day to meet somewhere for lunch. Please call me if you want to join us. I have some pamphlets and other information from the APF and anyone in the Denver area can call me if they want a copy of any of the items I have. Thank You to Desiree, Lelia, Wendy, Dr. Pierach and everyone who attended the gathering. Suzette Cowles Dsuzette41@msn.com

Wisconsin and Illinois Meeting This was my first IN TOUCH meeting, so I did not know what to expect. I was so surprised when fourteen people came, including patients, family members and professionals, all of whom wanted to learn about the porphyrias. We all shared our stories and found it fun to learn other people's experiences. One of the people had not been diagnosed yet, but since our meeting her test results came back positive. We watched the new DVD and really enjoyed it. Some of

the people wanted to purchase the DVD at the meeting, but I directed them to the APF website www.porphyrifoundation.com to buy a copy. (Editors note: The DVD is now available free with membership and can also be purchased via the APF office.)

The people who were there to support us meant a lot. They are also learning about this disorder and how it has changed our lives. It is a big change for everyone but if you look at it in a positive way it does get a little easier. And if we all stick together and keep talking, we will be a big help to each other.

God Bless, Jennie Eberhardt, Racine, Wisconsin

Georgia Meeting Of the twenty six people who came to the Georgia INTOUCH meeting at the home of Lelia Brougher, there were two sets of twins, a young woman who had just recovered from cancer surgery and was on crutches, six people who had driven more than two hours, a man whose case was published in a major medical Journal in 1968 and several men and women who had never met another person with porphyria. Lelia, who is the national IN TOUCH NETWORK coordinator, was pleased that the meeting was such a successful event. Desiree Lyon also attended the meeting, because she was visiting Lelia, her daughter, at that time. She told the group about the great new expanded physician education programs, including the CME course.

However, the attendees were enjoying such delightful camaraderie sharing their experiences that Desiree saved the remaining agenda for the next meeting. The group formulated several excellent suggestions, including the need for a program for the family members who manage the care of their loved ones when they are ill. Their physical and emotional needs are innumerable and often ill defined, so the burden on the caretaker can also very difficult, too. Your suggestions for this area are also needed.



Twins: Stephanie, Danielle, Tennile and Tiffany

Does Rose's Story Sound Familiar?

Rose is a beautiful young woman. She is 33 years old, has AIP and is an active member of the APF. Her first major attack was in the late nineties, which ended in misdiagnosis. Even though AIP was in her family, her physicians at Stanford decided that her test levels were not high enough to indicate porphyria. At the time, she was in terrible pain, could not walk and was experiencing other severe symptoms of AIP. Because her attack was near her menstrual cycle, the doctors eventually sent her to a gynecologist, which, of course, did nothing to solve her illness. So despite having AIP in the family, Rose assumed she did not have the disease and pressed on to find another answer.

The same scenario occurred again and again. Her misdiagnosis continued to cause her to be without proper medical care. Finally, Rose decided to take matters into her own hands, because by that time, she believed that she had the porphyria regardless of what she had been told by the medical community. Thus, when she experienced her next attack, she placed her urine in the window as she had read to do and watched it change to a darkish brown color over the course of the day. To test her theory, she also placed her husband's urine in the window sill as well, but it remained the same color. (*Editor's note: This can occur but does not always happen, so it should not be used as a diagnostic technique.*) Armed with this piece of convincing evidence, she was sure that she had AIP. She contacted a new doctor and explained her plight. The new doctor promptly reached out for help and contacted Dr. Karl Anderson, a porphyria specialist in Galveston, Texas. That was the turning point for Rose. Dr. Anderson asked that a series of tests be sent to his porphyria lab, and after a few days, he called with the answer; Rose had AIP. After all the years of misdiagnosis and sickness, she had her long awaited answer.

Rose then entered into two porphyria research projects, because she wanted to play a part in finding a treatment or cure for herself and others who were suffering as well. One was with tin mesoporphyrin and the other was lupron., but her best results were derived when she was administered Panhematin. When she experienced her next attack, she had relocated and had a new physician. He did not call Dr. Anderson for his advice as he had promised to do and did not give her Panhematin, so sadly, her attack became life threatening. Rose was in such critical condition that she does not remember much of that attack; her brain was swollen, she was hallucinating, and her electrolytes were badly imbalanced. Finally, almost a month later, she was given Panhematin, and she began to improve.

However, since Rose had still not recovered completely, she decided to travel to Dr. Anderson for treatment. He placed her on Panhematin, and her attack subsided completely. Unfortunately, later when she moved to San Jose, she suffered yet another attack, but she could not convince the doctor to prescribe Panhematin. The result was a lingering attack. She then found a doctor who would prescribe Panhematin. When she had her next attacks, she was given Panhematin early in the attack as indicated, and the attack subsided quickly. Fortunately, she has remained well since her last infusions and is living a normal life. She has even just returned from a vacation in Hawaii. "It is so wonderful to be without pain and not be sick. I haven't felt this way in four years. In fact, I had forgotten what it feels like to feel this well."

Keeping IN TOUCH through the APF IN TOUCH NETWORK

Because the list of APF members who want to be a part of the *IN TOUCH NETWORK* has grown so large, we can no longer post them in the newsletter. Therefore, if you would like to find other APF member of the *IN TOUCH NETWORK* in your area, please contact the APF office. We maintain the *IN TOUCH NETWORK* database and update it daily. We can connect you with other members who live nearby. If you would like to participate in the *IN TOUCH NETWORK*, please contact the office, and we will then send you a consent form. It is important to have permission from each of you to give your name and contact information to other APF members. Keeping in touch with other people with porphyria has been lifesaving to many of our members and to others it has been a wonderful means of finding new lifetime friends. You will benefit greatly by keeping IN TOUCH. The first step is to contact the APF office, and the next is to enjoy your new friends. The *IN TOUCH NETWORK* has been a great resource for learning about porphyria and sharing your own experiences.

Join the APF or update your membership dues and receive the *Porphyria Live DVD* FREE



Chicago Digestive Disease Week

When Desiree asked me to man the exhibit booth at the Digestive Disease Week (DDW) international medical convention, I felt it was a true honor to serve. I have only been diagnosed for a year and a half, so I have much to learn. I had to make decisions about

Jennie Eberhardt and Dr. John Hughes from Houston

this as I have not traveled in over two years due to my health and all the paraphernalia I have to bring with me. But when I thought of what I could give and learn, all of the other issues did not matter. The most important thing for me was to meet and talk with many of the 10,000 medical professionals from around the globe whose specialties were digestive diseases.

My husband, Richard, went with me to make sure I was going to be OK and to help me set up and man the APF exhibit booth. When we settled into the booth at the beginning of the DDW, Richard was talking with the doctors like a pro. I discovered that he had completed reading the books and literature before me. It was a reminder that our family goes through a lot and are trying to help and understand what has happened to me. I was reminded to say how much I appreciate him.

This learning experience was unbelievable. The medical professionals were very willing to talk and receive information. I was glad I had a rash on my hands, so they could see the porphyria lesions. Most of the doctors also enjoyed hearing about porphyria from a patient's perspective. It was especially wonderful to see doctors from my home town, who had never seen a case of porphyria. I told them all that it took 48 years to diagnose me. It would have made my life easier to have been diagnosed earlier. Some of the doctors even told Richard and me stories about their porphyria patients. They were glad to know about the APF and their vast source of information.

The entire experience was so worthwhile. Not only because of the part we could play to educate the doctors, but also to learn what occurs at the APF. There is too much to list that

transpires on a daily basis at the APF and that some of the volunteers are sick like some of us. I do hope I can volunteer again for another challenge like the DDW. The whole experience made me feel worthwhile in a way that I haven't felt for two years. It also showed me I have so much left to learn and many questions to ask. If I am asked, I would be proud to participate again, because this has been a life changing experience that I will never forget. I met fantastic people, including Dr. Hughes from Houston, who asked me to come speak to other doctors. It was the opportunity of a lifetime!!!! By Jennie Eberhardt, Racine, Wisconsin

Be a Volunteer at a Medical Convention

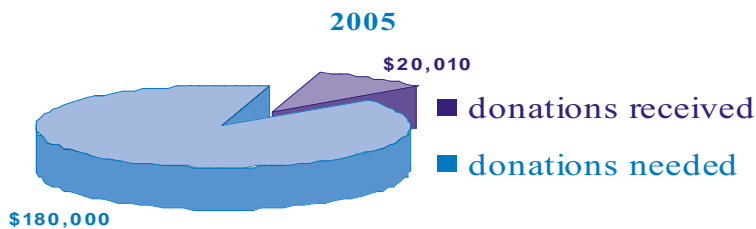
You, too, can participate as a volunteer at a medical convention. Later this year, we will be exhibiting at two more major conventions; *The American Academy for the Study of Liver Diseases* in San Francisco Nov.11-15 and the *American Society of Hematology* in New Orleans Dec. 3-6. These are the two important medical conventions for physicians will diagnose people with the acute porphyrias. We also exhibit at the *Academy of Dermatology* to educate doctors who diagnose and treat the cutaneous porphyrias. Your help is needed to man the booth. If you would like to volunteer, please contact the APF for more information.

Continued (Lyon's Share...) from page 1

Unfortunately, some volunteers did not understand why they were not chosen even though they had graciously volunteered. As discussed, the researchers cannot enlist people who do not fit the protocol. Rather, they must adhere to the set strict guidelines. We appreciate every person who has volunteered as a research participant, whether you became involved or not. The fact that you were willing to offer your services is a noble gesture. Hopefully, there will be a different project in the future that will need participants, and you will be needed again.

Continued (The Heme Pool) from page 2

work and restore normal operations. If only he would have known to stay away from those irritants, he may not have had any problems! *This article is compliments of our friends at The Canadian Porphyria Foundation!*



"Protecting Our Future" Update

The opportunity of a lifetime has come our way!

We can make a dream come true!

With your help, we have the opportunity to secure one of the brightest stars in the medical field as a porphyria specialist and developing a major porphyria center. To do this we will need to recruit him to an existing porphyria center, all of which will cost approximately \$90,000 a year. Such an opportunity is rare, thus reacting to it timely is a necessity and at present as you can see, we are far from our goal. However, I know that together you and I can raise the funds needed to accomplish our most important goal. One generous woman, who did not even suffer with porphyria, contributed the largest donation we have received to help train young specialists. We thank her for her generosity!

The APF notified our members about our "Protecting the Future" campaign. The mission of this campaign is to attract and train the next generation of doctors and future specialists in the field of porphyria. Over the next decade, we all will lose ninety percent of our porphyria experts, who have led the field of porphyria research, testing and treatment for the past thirty years. At present, we have only a handful of experts now. Without financial support, we run the risk of losing their expert knowledge of the disease, quality testing, diagnosis, treatment and ultimately research for a cure. Fortunately, we have been handed a silver platter opportunity to recruit a well renowned, young doctor with extensive experience, and interest in training others in the porphyrias.

Please send your donation to our "Protecting the Future" fund in the enclosed envelope to help secure a healthy future or ourselves and our children. For questions, call our campaign coordinator, Lelia Brougher at 404-550-4880 email@broughers.com or Desiree Lyon at the APF: 713-266-9617 or email lyonapf@aol.com.

A New Brochure

The APF has published a new 15 page brochure, **Management of the Acute Porphyrias**, which contains very comprehensive information on the diagnosis and treatment for the AIP, VP, HCP and ALAD. You can purchase this brochure for \$1.00 each.

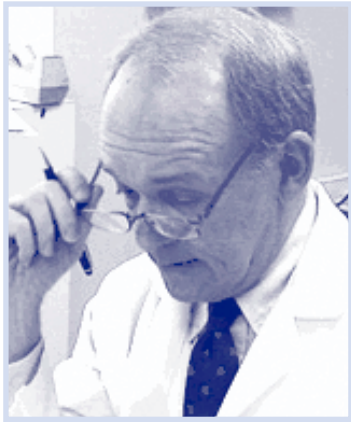
You might consider procuring a supply of these brochures or each of your and to keep on hand for medical professionals who might treat you in the emergency room or other urgent care facilities. Everyone with the acute porphyrias should carry one of these brochures along with their Medic Alert emblems. To order the brochures; email Elizabeth at Elizabethapf@aol.com or phone the APF: 713-266-9617.

Management Sequence-Acute Attacks

As part of the APF expanded physician education program, we have published a new physician brochure for the diagnosis and treatment of the acute porphyrias, **Managing the Acute Porphyrias**. In addition, we also hope to publish articles in the upcoming newsletter to assist physicians in managing your health care. Like the one below. The following is a sample guideline for managing acute attacks.

- 1) Document substantial increase in urine PBG or ALA and collect samples to determine the type of acute porphyria.
- 2) Symptomatic supportive treatment and monitoring to include:
 - Check Serum electrolytes, creatinine, Mg at least daily
 - Provide intravenous fluids for dehydration and correct electrolyte imbalances
 - Give Intravenous glucose (10%) at least 300 grams daily until Panhematin is available
 - Provide appropriate pain and nausea medications
 - Prescribe Beta Blocker for hypertension and tachycardia if clinically indicated
 - Use Intensive Care unit if respiration is impaired, monitor vital capacity
 - Instate seizure precautions, especially if hyponatremic
 - Avoid medications not known to be safe with porphyria
 - Monitor bladders distension and catheterize if necessary
 - Hold oral feeding if nausea, vomiting or ileus is present
 - Provide intravenous nutrition support if oral intake not possible after 1-2 days
 - Manage central line if present to avoid infection
 - Monitor neurological status, including muscle strength

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Porphyria Specialist: Dr. James Kushner

As part of our expanded physician education program, we will be profiling porphyria experts in upcoming newsletters. We are very proud to have such distinguished physicians working with us for almost three

decades. **Dr. James Kushner**, our first profile, is renowned in the field of porphyria and hematology and has shared his expertise with many primary care doctors who needed assistance in diagnosing and managing your health care. Dr. Kushner also participated in the publication of the recent *Annals of Internal Medicine*.

Dr Kushner has served for more than thirty years on the faculty of the University of Utah School of Medicine, where he currently holds several positions including the Maxwell M. Wintrobe Distinguished Professor of Medicine, Professor of Medicine in the Division of Hematology/Oncology, Program Director of the General Clinical Research Center, and Chief of the Division of Hematology in the Department of Internal Medicine. Dr. Kushner is also a member of the Advisory Council for the Center for Scientific Review at the National Institutes of Health.

He has authored over one hundred articles published in various major medical and scientific journals, including publications on the porphyrias. He has an active hematology and porphyria research program funded by several awards from the National Institutes of Health. Dr. Kushner received his M.D. degree from the University of Pittsburgh School of Medicine and a B.A. from Bowdoin College. In addition, Dr. Kushner has also developed and led the Clinical Research Curriculum to provide clinical researchers with the necessary and appropriate background for successful clinical research. His research has greatly enhanced the understanding and treatment of the acute porphyrias, as well as Porphyria Cutanea Tarda. Dr. Kushner has also been instrumental in the treatment of CEP patients, including little Nicholas Ashby, who has most recently endured a bone marrow transplant for CEP.

Transdermal Estrogen Replacement Therapy in Postmenopausal Women Previously Treated for Porphyria Cutanea Tarda

Dr. James Kushner and his colleagues at the University of Utah published a paper on the subject with the above title. The focus was oral contraceptives and postmenopausal estrogen replacement therapy, which are recognized as risk factors for the development of porphyria cutanea tarda (PCT) in women. The recommended clinical practice is to withhold estrogen therapy in women who have had phlebotomy therapy for PCT and are clinically and biochemically normal. They tested the safety and efficacy of transdermal estrogen replacement therapy in 7 women previously treated for PCT and compared them with 19 non-porphyrin control subjects treated with transdermal or oral estrogens. Gonadotrophic hormone levels, estrogen levels, liver function studies, body iron stores, urine porphyrin excretion, and cytochrome P4501A2 (CYP1A2) activity were monitored for 1 year.

Four of the women previously treated for PCT completed the study, and none had evidence of a porphyric relapse. Gonadotrophic hormone levels fell and estrogen levels rose in all women receiving estrogens. The administration of estrogens by the transdermal route appeared to be safe in the small number of subjects studied. If you had had difficulty with these patches, please contact the APF.

Continued (Management Sequence...) from page 6

3) Specific Treatment Intravenous hemin 3-4 mg/kg body weight once daily for 4 days. Start as soon as possible for all but mild attacks and extend treatments as clinically indicated.

4) During Recovery

- Complete molecular studies to identify mutations
- Plan rehabilitation program for recovery if severe paresis is present
- Educate regarding safe drugs and diet
- Consider other preventive measures, based on contributors to the attack. e.g. GnRH analogues for preventing frequent recurrent cyclical attacks

You Can Help—Danielle Frazzini, is planning an annual Porphyria Race to be held in Atlanta. She needs your help to make this undertaking a success. Also, if you work for a company that might lend their support or if you can participate, please contact Danielle at dfrazzini@yahoo.com.



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

Free Continuing Medical Education Course for physicians can be accessed on the APF website.

Chat Room find out the time and place on the APF [web site: www.porphyrifoundation.com](http://www.porphyrifoundation.com).

The new Acute Porphyria Brochure can be purchased for \$1.00 each.

To Purchase the ER Kit, DVD, Book, contact the APF.

The Emergency Room Kit and the DVD *Porphyria Live* can be purchased for \$20.00 each.

The book, *Porphyria A Lyon's Share of Trouble*, can be purchased for \$20.00.

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