AMERICAN PORPHYRIA FOUNDATION

3rd Quarter, 2007

Exciting New Research Project: Dr. Herbert Bonkovsky

Life requires energy. Except for mature red blood cells, all cells in the body have organelles called "mitochondria", which are the "energy plants" of cells. The carbon and hydrogen and other burnable elements that are contained in foods are 'burned' or oxidized in our cells, especially in mitochondria, which combine hydrogen with oxygen, leading to the formation of water and the production of large amounts of energy. Part of this energy is released as heat, but, normally, most is conserved and accumulated in high energy compound called ATP. ATP provides chemical energy for virtually all of our energy needs. In order to utilize the energy stored in hydrogen, mitochondria split hydrogen atoms into electrons (e-) and protons (H+), which are carried step by step along a chain of heme-containing enzymes called cytochromes to oxygen. This is where porphyrins come into play. Porphyrins are necessary to construct a complex molecule, called heme. In heme, a porphyrin ring, composed of four pyrroles strung together and closed back on itself, hold an atom of a iron in its center. These atoms of divalent metals can take and give electrons. Therefore, heme structures are important not only for mitochondrial enzymes, but also for many other biological processes. This is how, for example, hemoglobin in red blood cells can carry oxygen, and how electrons are carried in mitochondria from hydrogen to oxygen. This is why disruption of heme formation has dire consequences for virtually all organs – it damages the supply of energy to the cells. Commonly, the disruption of heme formation is associated with abnormal metabolism of porphyrins.

The acute porphyric syndrome may occur in several hereditary diseases or inborn errors of metabolism. These include the so called acute or inducible porphyrias, namely, acute intermittent porphyria, hereditary coproporphyria, variegate porphyria, and the rare form of porphyria due to severe deficiency of 5-aminolevulinic acid (ALA) dehydratase (also called ALA dehydratase deficiency porphyria). In addition, similar syndromes may occur in hereditary tyrosinemia and the acquired disorder of lead poisoning. The underlying causes of these syndromes are deficiencies in the normal formation of porphyrins and heme.

The nervous system (NS), composed of the brain, the brain stem, the spinal cord, and the nerves that go out from or report back into these structures, is one of the most sensitive to heme dysfunctions because neuronal cells require a very large quantity of energy. In our laboratory we have developed a number of highly sensitive quantitative methods, which allowed us to determine and localize precisely the sites where mitochondrial dysfunction occurs in such neurodegenerative diseases as Huntington's disease, Parkinson's disease, and Lou Gehrig's disease [amyotrophic lateral sclerosis]. In this new project that we hope will be supported by contributions from the APF and its members and friends, we will bring our methods and experience to bear upon analyses of mitochondrial dysfunctions, to determine mechanisms by which neuronal cells may be damaged in patients with acute porphyria. Because mitochondria are so important for our survival, they are the sites where the fate of cells is determined. It was discovered recently that during cell stress and damage, different 'death signals' are translated to mitochondria in the form of increases in calcium ions. How well or badly mitochondria can handle these increases determine whether the cells will die or live. We have developed methods to determine how well mitochondria can resist to the death signals, by determining quantitatively how much calcium mitochondria can sequester.

In addition, during mitochondrial respiration, some electrons can "jump" onto oxygen molecules. As a result, a free radical oxygen molecule is formed, called "superoxide radical". From this radical other forms of oxygen-containing radicals may form, collectively called "Reactive Oxygen Species" (ROS). Because these radicals are very active chemically, they damage many components of cells: nucleic acids, proteins, lipids. It is believed that these radicals are responsible for the process of aging, because "errors" are accumulated and cells become less viable or turn into cancer cells. We use highly sensitive method to evaluate quantitatively formation of ROS by mitochondria. We believe that our appraisal of selected mitochondrial functions, including ATP synthesis, calcium ion retention, and generation of ROS, will allow us to determine mechanisms by which toxic metabolites and/or heme deficiency, which may occur in acute porphyria, disrupt energy metabolism in brain and spinal cord mitochondria. Gaining such knowledge is the first step along the path to preventing and treating the acute porphyrias better.

Editor's note: The costs of the reagents and supplies for a year are $15,000. This does not even include a laboratory technician to assist Dr. Bonkovsky. Your help is needed. Such a research project is extremely costly and government funding for rare diseases is almost non-existent. This is our opportunity to make this project possible through your donations.

RESEARCH IS THE KEY TO YOUR CURE

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Dr. Brendan McGuire Congratulations

We are proud to announce that Dr. Brendan McGuire has been selected as the newest ***Protect the Future*** candidate. Dr. McGuire is the Medical Director of the University of Alabama Liver Transplantation in Birmingham, Alabama (UAB) where he also serves as Associate Professor of Medicine. He will be training as a porphyria expert with Dr. Joseph Bloomer, who has been renown in the field for over three decades.

He received his Bachelor of Science in Chemical Engineering at the University of Notre Dame, his Masters of Science in Biochemical Engineering at Pennsylvania State and his Doctorate in Medicine at the University of Pittsburgh.

Board Certified in Gastroenterology and Internal Medicine, Dr. McGuire's primary focus is in the clinical management of complications in patients with end-stage liver disease at the University of Alabama. He is the principal of the acute liver failure study group and a pilot study looking at the prevalence of kidney pathology in patients with hepatitis C cirrhosis undergoing liver transplantation at UAB. In addition, he is interested in the effects of hepatic encephalopathy on quality of life, to investigate new drugs to treat hepatic encephalopathy in patients with cirrhosis, and to identify factors to predict a successful outcome after liver transplantation. His long term goals are to expand the UAB center of excellence into a leading national center for clinical care and research in patients with end-stage liver disease.

Along with his research commitments and teaching responsibilities, Dr. McGuire is also widely published in major medical journals and medical textbooks. We are proud that a physician of such distinction has also taken on the challenge of becoming one of the leading experts in the field of porphyria.

Your donations are needed to help fund the training of more porphyria experts through the ***Protect the Future*** *program. This program is vital to your future health. Please use the enclosed envelope for your donation and mark it* ***PTF****. Thank you.*

Porphyria Centers

The APF promotes comprehensive care necessary for treating individuals with porphyria. We put patients and their doctors in touch with specialists, laboratories and Porphyria Medical Centers that provide the highest level of knowledge to treat porphyria. Porphyria Medical Centers are located in cities across the United States. The specialists at these centers have been treating porphyria patients for decades and are highly published in the field.

In comparison to more well-known diseases, physicians with expertise in porphyria are fewer in number and are scattered around the U.S. Oftentimes, this makes locating a knowledgeable doctor in your city improbable, if not impossible. If you need assistance locating one of these Porphyria Centers, please contact the APF at 1-866-APF-3635.

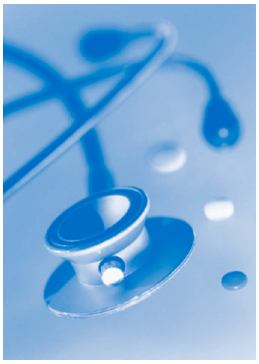
EPP INFO

The following article was published in the EPPREF news. It answered many of the questions we have received from our members with EPP. We thank the EPPREF news and its publisher, Dr Micheline Mathews Roth, for permission to reprint this excellent article:

Gene Therapy Update: In EPPREF NEWS issue Number 35, we discussed gene therapy for EPP, and mentioned that the Roth lab team collaborating with scientists from the Harvard-MIT Health Sciences and Technology group had cured the mouse model of EPP with gene therapy targeting the bone marrow. Our group was the first to do that, publishing our results in the July, 1999 issue of the journal, "Nature Medicine". Since our results were published, other scientists have also cured the EPP mouse model with bone-marrow gene therapy using different viral vectors than we did. However, as was reported at this year's meeting of the American Society of Gene Therapy, there are still some safety issues with viral vectors and bone-marrow gene therapy in people, as we had reported there were problems in issue 35. Although progress has been made in understanding and correcting the problems, they are not all solved at this time. Thus, unfortunately, we are not quite yet ready to attempt gene therapy in EPP people.

Sunscreens: *Since summer is still not over, we are also including the sunscreen information from the EPPREF.* According to data from its makers, Mexoryl SX only works well for short-wavelength UVA rays – those from 320 to 340 nanometers. The rays that bother EPP people are those from about 360 to 500 nanometers, in the visible range of the solar spectrum. However, if Mexoryl is in a sunscreen that also contains some blocking agents, such as zinc oxide or titanium dioxide, that sunscreen might be worth trying, as the blocking agents will be what will be effective for EPP people.

Another EPP person mentioned Sephora (www.Sephora.com) as another company that markets sunscreens. I checked their listings, and two sunscreens they list may work for EPP people: 1) Korres Natural Product Watermelon Sunscreen Face Cream – it contains titanium dioxide and zinc oxide, and 2) Bliss Oil-free Sunban Face cream – it contains titanium dioxide. Although these products are both listed as face creams, there should be no reason why they would not also work on arms or hands. We have mentioned in the past COTZ, which is made by Total Block (www.totalblock.com) – it contains zinc oxide and titanium dioxide, and can be used over the entire body. However, please remember our past suggestions about trying a new sunscreen – just put it on a small area, such as part of an arm, and expose that area, while keeping the rest of your body covered as you usually do to prevent reactions. Keep the area on which you have applied sunscreen exposed for the time it would take for you to get your symptoms, then cover it, and see if you get a reaction. Don't forget to let us know if you find a sunscreen that prevents your symptoms – it may work for others too!



Dietary Supplements: Condition Worsens

Critics of the dietary supplement industry say the industry is still operating without FDA oversight despite evidence of substandard quality and safety problems. The influx of ingredients from China has made the problems worse in recent years, because the Chinese government does not test the exports and has few rules governing manufacturers. In what appears to be retaliation, the Chinese government returned or destroyed certain US nutritional supplements, saying they failed to meet Chinese safety standards. Nonetheless, it has been suggested by porphyria expert that these Chinese supplements and treatments should not be taken since they have not been investigated for safety in porphyria.

Tips for Doctor-Patient Communication
Before Your Doctor Visit

Prepare a concise list of all of your medications, refills, a summary of your medical history, questions, concerns and symptoms. Complete your tests before your visit to help your doctor better determine your treatment during your visit.

At Your Visit

Be on time. Be as brief and concise as possible as you discuss your lists and questions. Your doctor may only have 15 minutes to spend with you. If you do not understand the doctor's conversations, tell him/her and ask to review the issues. Factors can confuse communication, namely, emotions, communication style, different goals and lack of time all work against us. When emotions are high, logic is low. Remember that major medical journal articles are far more accepted than internet articles.

After the Visit

Often patients have questions they forgot to ask. First, check the educational materials to see if the question can be answered there. If it is urgent, call the office right away and note that it is urgent. If not, ask that the doctor call you and then. It is best to have your questions clearly written. Be aware that the doctor may not be able to answer your call until the end of the day, but a nurse or physician's assistant can usually help earlier.

Welcome to "SAKURA Friends"

The Japanese porphyria patients group, "SAKURA Friends," (SAKURA means cherry blossoms) was formed in 1997 by porphyria patients, families and support volunteers. Today the group has 66 members who work together to enhance knowledge, research, diagnosis and treatment for the porphyria while encouraging communication and support among members. They publish three newsletters annually and organize gatherings among the members

Our friends in Japan are eager to make friends outside Japan to exchange information and experiences and learn from each other.

Those who read Japanese can find more information about their group at: www.sakuratomonokai.com.

Mexico: A Global Partner

Susana Isla is our contact for Mexico, our new Global Partner. We welcome her and thank her for volunteering to assist us with our partner support program. If you would like to communicate with Susana, contact the APF.

My name is **Susana Isla**, and I live in Mexico. From the time I was five years old until I was forty-two, I have had a severe itching that never stopped. I thought it was psoriasis. After I learned about porphyria, I realized that it was not psoriasis. Aside from horrible itching, it also bled and was scaly, particularly after I went to the beach or some sunny activity. My skin looked as if I had leprosy and was very painful all the time. It was so intolerable that during the night I would have to take off all my clothes. I have never been able to tolerate heat on my body like a normal person, but I did not know why and neither did the doctors.

I also suffered insomnia, so I went to a sleeping clinic. No one at the clinic knew what was wrong either. I also had constipation so severe that I endured major bowel obstruction. In addition, I was hospitalized several times after I drank alcohol. These were all clues that might point to a porphyria diagnosis. I had repeated tests, including blood tests, but none of my doctors thought about porphyria. Regardless of the many tests, the doctors could not come up with an answer. This is what happened to me even among specialists until one doctor finally discovered I had porphyria.

Thanks a lot APF for caring and for helping people with this disease. I am not yet too knowledgeable. It is so new to me, but I will study and try to help others learn about it. I have even contacted every one that has treated me so that when they have a patient like me they will know what to do instead of sending them back home without a diagnosis and the wrong medicine. I will be glad to start raising the consciousness about porphyria in Mexico.

Read an expanded version of Susana's story in Spanish in the Global Partners section of the website.

Family Health Portrait

The *My Family Health Portrait* allows you to create a personalized family health history report from any computer with an Internet connection and an up-to-date Web browser. Information you can provide creates a drawing of your family tree and a chart of your family health history. Both the chart and the drawing can be printed and shared with your family members or your healthcare professional. Used in consultation with your healthcare professional, your family health history can help you review your family's health history and develop disease prevention strategies that are right for you, which is particularly helpful for those with porphyria. See http://familyhistory.hhs.gov/.

Mt. Sinai Porphyria Center Opens

In our last newsletter, we announced that the Mt. Sinai Medical Center in New York City was opening a **Porphyria Center** where patients can be tested and treated. Longtime APF member, **Patsy Brady**, (left) was the first patient to visit the clinic. Despite the fact that NYC is one of the world's largest cities, the many patients who live in the area have been without a porphyria clinic for almost 20 years. Dr. Kenneth Astrin, who coordinated all the activities of the DNA lab and clinic with us, has left Mt. Sinai (see article on page 7). Dana Doheny, the genetic counselor for the *Protect the Future Program*, will communicate with the patients, as Dr. Astrin's did so kindly and adeptly, while Victoria Njoku will coordinate the porphyria clinic and appointments.

Without a specialist to oversee the center, however, patients had no one to diagnose or treat them. Several of our members responded to this serious need and provided part of the funding to open the center. Now we need your help to open other centers around the country and train more experts through our **Protect the Future Program**. Lelia Brougher represented the APF at the opening of the center and personally thanked Dr. Manisha Balwani and Dr. Lawrene Lui for undertaking the training with porphyria experts to gain the expertise about porphyria to maintain the center. However, we cannot open centers and train doctors without your help. We have been making tremendous inroads in these projects and together we can achieve what we need: better healthcare for porphyria patients. Below are photos from this exciting event.

If you are interested in making an appointment with Dr. Balwani or Dr. Lui at Mt. Sinai, please call Victoria Njoku at 212-241-6790.

Elizabeth having the DNA swab

Dr. Astrin, Dr. Lui, Lelia, Dr. Balwani, Dana

The Grand Opening

Enclosed is an envelope for your financial support, your questions, and your concerns. Thank You.

IN MEMORY: After the passing of their son, Joshua, Nor San Juan writes, "Let me take this opportunity to thank all of your for the prayers, care/concern, financial support and medical advice." Also, Joe Dyer was recently killed in a terrible car wreck. Joe and his late wife, Betty, were very early members of the APF and assisted us at the San Francisco medical conventions for decades. We are saddened by the passing of our many friends and express our sympathy to their families. The following people also made generous gifts in MEMORY of their loved ones. We sincerely wish to express our sympathy to the families and thank them for their memorial tributes to the APF.

Elaine Smuczynski for ***James Smuczynsky***
Donna Pagano for ***Arlene Yager***
Sophie & Edward Marshall for ***James Smuczynski***
Desiree Lyon for ***Dr. Richard D. Levere***
Sydelle E. Diner for ***Dr. Richard D. Levere***
Lond & Ernie Hyne for ***Marcia Taylor***
Vickie Gehm for ***Judy Coley***
Marianne Danko for ***Helen Smuczynski***
Marianne Danko for ***James Smuczynski***
Carol & Arthur Herring for ***Birtie M. Prine***
Jo Ann Martin for ***Birtie M. Prine***
Jason Winters for ***Birtie M. Prine***
Gloria and Dave Morgan for ***Birtie M. Prine***
Donna L. Pagano for ***Arlene Yager***
Donna L. Pagano for ***Millie O'Toole***
Desiree Lyon for ***Joe Dyer***

IN HONOR: We also received donations from families who wished to honor their loved ones. We also thank them for their generous gifts.

Sara Tamburrino for ***Shelly Hill***
Sharon I. Koch for ***Debbie Koch***
Carl Tishler for ***Dr. Peter Tishler***

Guide to Drug Porphyrogenicity Prediction and Drug Prescription in Acute Porphyrias

Doctors Stig Thunell, Erik Pomp, and Atle Brun from the Porphyria Center at the, Karolinska University Hospital in Stockholm, Sweden and the Norwegian Porphyria Centre, at the Haukeland University Hospital in Bergen, Norway and the Institute of Medicine at the University of Bergen, Norway, recently published an important article, *Guide to Drug Porphyrogenicity Prediction and Drug Prescription in the Acute Porphyrias* in the ***British Journal of Clinical Pharmacology*** (See: DOI:10.1046/j.0306-5251.2007.02955.x)

Their paper addresses two of the most vital issues for people with acute intermittent porphyria (AIP), variegate porphyria (VP), hereditary coproporphyria (HCP) and ALAD-deficiency porphyria (ADP) or are carries of the diseases. The choice of safe drugs and the strategy to use when potentially unsafe drugs cannot be avoided is difficult. The drug databases for acute porphyria are meant to be used by and discussed with health care professionals. Further advice should be obtained.

For over a hundred years, therapeutic drugs have been known to precipitate acute attacks. Since that time, lists of potentially safe or unsafe drugs have been developed. However, drug lists for acute porphyrias are largely based on anecdotal evidence, thus there are discrepancies between lists. These lists are of tremendous benefit and have contributed significantly to the reductions in unnecessary illness and deaths. However, because there is an incomplete basis for prediction of the porphyrogenic potential of the drugs, has often prompted the application of a 'better safe than sorry' strategy in classification work. Thus, under-medication has occurred when physicians are not able to determine if a drug is safe or not.

As quoted in their article, the investigators present, "a technique for prediction of risk that a certain drug may activate the disease in a gene carrier for acute porphyria. Their premise is based on a model explaining the clinical manifestations as a result of the acute overloading of a deficient enzyme within the hepatic heme biosynthetic chain." About 1000 therapeutic drugs were categorized with regard to porphyrogenicity by the technique proposed and are in the *Links* section in the APF website.

Congratulations Lauren Warren

And thanks for the $3000 she raised for the APF.

Well, it's official, I am an Ironman! The race was Fantastic! What an incredible experience Over 3,000 volunteers for 2,400 athletes and the day was flawless! Even more incredible news is that on my first try ever at Ironman distance I finished in 4th place and qualified for the ***Ironman World Championship*** in Kona, Hawaii!! We are still in shock over since national and international athletes compete at Ironman Lake Placid to qualify for the World Championship in Hawaii. Many athletes try for years and never qualify, so we are still in shock.

Two days before the race I had an attack, but quickly got it under control with very careful food intake etc., but it did greatly scare me that it would put me out of the race if I didn't. God is good! Morning temperatures were chilly at 48 degrees but quickly warmed up and reached the upper 70's by race end, all in all, picture perfect weather. I had one of the fastest swims in the competition. The bike was beautiful and I was able to carry out my nutrition as planned during all of my training rides. The run was perfect. For 26 miles they had an aid station every single mile loaded with orange slices, grapes, watermelon, pretzels, chicken broth, gels, Gatorade, water, and Coca-Cola.

The finish was a dream come true. I felt a tired, but strong. They escorted me into the medical tent after the race and weighed me, I had lost 5 pounds – pretty typical, nothing that a few days of re-hydrating and eating wouldn't help. There was a local Christian church that set up aid stations every mile distributing drinks, food and prayers for those, like me, who wanted them, I was thinking about everyone who has Porphyria and all the prayers that were going on during this race and the strength I received from it was incredible! When all was said and done, God saw me through to an incredibly fast finishing time, a 4th place finish, and a World Championship qualifying spot – absolutely amazing. Thank you all for being such a wonderful part of this journey.

God Bless, Lauren Warren

Dr. Peter Tishler, who oversaw the compilation of the APF drug safety list, is assisting us with a revitalized project. In the past, we have initiated a number of efforts to educate pharmacists about safe and unsafe drugs and the mechanism behind these lists. Until now, however, we have not had a major pharmacist organization behind us. Recently, Dr. Tishler contacted Ms. Cynthia Reilly, R.Ph., who is the Director of Clinical Standards and Quality Practice Standards and Quality Division of the American Society of Health-System Pharmacists (ASHSP) in Bethesda, Maryland. Through the ASHSP network, the APF can distribute the drug safety information, including the APF drug list website. This is a major resource to help educate about one of the major issues in the porphyrias. We sincerely appreciate Dr. Tishler and Ms. Reilly and thank them for their time and efforts in the most important project.

Another effective measure to educate pharmacists is for each one of you to educate your local pharmacist. Please take a copy of the drug section on the APF website or order a drug brochure for your pharmacist.

DNA and A Royal Research Project

Dr Alan Rushton, a distinguished pediatrician and medical geneticist from the Hunterdon Medical Center in New Jersey, is also a medical historian with a portfolio of many interesting articles, including written a history of medical genetics in America between 1800 and 1922. He says that his interest in the history of genetics was stimulated by professors at his undergraduate college, Earlham. After Earlham, he acquired the M.D. and Ph.D. (in genetics) at the University of Chicago, followed by residency training at Yale.

Recently, he began researching the relationship between the royals and individuals with porphyria now. Interestingly, we have discovered several ancestors of the British Royal Family and King George III through the APF newsletter, IN TOUCH and E-news. To further assist Dr. Rushton's interesting project, an APF member is supporting the DNA testing of two individuals, whose DNA would suggest a mutual inheritance and a VP diagnosis. If they both have the mutated VP gene and share the same mutation, that would argue for the VP gene being present in the British Royal Family from the Norman French line in the eleventh century. Watch for the exciting results.

Porphyria Around the World

Our mission is to help our friends around the globe enhance their own patient support organizations through education and our Global Partners Program:

* **Hepatoerythropoietic porphyria: a missense mutation in the *UROD* gene is associated with mild disease and an unusual porphyrin excretion pattern:** D.K.B. Armstrong, P.C. Sharpe, C.R. Chambers, S.D. Whatley, A.G. Roberts and G.H. Elder, **BRITISH** Journal of Dermatology, vo l 151 Issue 4,, Oct. 2004 Prevalence of Acute Intermittent
* **Porphyria in a MEXICAN Psychiatric Population:** Jara-Prado A, Yescas P.; Sanchez F.J.; Ros C.; Garnica R.; Alonso E., Archives of Medical Research, Vol.31, Jul 2000,
* **Acute intermittent porphyria associated with hyperthyroidism**, Vithian, Karvnakaran; Samat, Ashish; Jones, M Keston, Annals of Clinical Biochemistry **INDIA** Volume 43, Number 5, Sept 2006
* **Treatment and Treatment Considerations in a Patient with Advanced Breast Cancer and Acute Intermittent Porphyria: SWEDEN** Kristiansen, Charlotte; Tyge Langkjer, Sven Acta Oncologica, Volume 45, April 2006,
* **Dual porphyria with mutations in both the *UROD* and *HMBS* genes** Harraway, James R.; Florkowski, Christopher M.; Sies, Christiaan; George, Peter M. **ENGLAND** Annals of Clinical Biochemistry, Volume 43, Number 1, January 2006, pp. 80-82(3)
* **Characterization of two isoalleles and three mutations in both isoforms of purified recombinant human porphobilinogen deaminase** Brøns-Poulsen, J.; Christiansen, L.; Petersen, N. E.; Hørder, M.; Kristiansen, K. **SCANDINAVIAN** Journal of Clinical and Laboratory Investigation, Volume 65, Number 2
* **Anesthesia in a child with homozygous porphobilinogen deaminase deficiency: a severe form of acute intermittent porphyria** Sheppard, Dorman, **ENGLAND** Pedriatric Anaesthesia, Vol 15,May 2005
* **Novel HMBS founder mutation and significant intronic polymorphism in SPAINISH patients with acute intermittent porphyria:** E. Guillén-Navarro; P. Carbonell; G. Glover; M. Sánchez-Solís; A. Fernández-Barreiro Annals of Human Genetics, Volume 68, Number 5, September 2004, pp. 509-514(6)
* **Pro-oxidant and antioxidant factors in acute intermittent porphyria: Family studies ITALY** Rocchi E.; Ventura P, RoN-zoni A.; Rosa M.C.; Gozzi C.; Marri L.; Casalgrandi G.; Cappellini M.D., Journal of Inherited Metabolic Disease, Volume 27, Number 2, 2004, pp. 251-266
* **International air travel: a risk factor for attacks in acute intermittent porphyria** Authors: Peters T.J.; Deacon A.C., Clinica Chimica Acta **ENGLAND**, Volume 335, Number 1, September 2003
* **Safety of fluoxetine treatment in a case of acute intermittent porphyria**: Si·bel Mercan; Oguz Karamustafalio-glu; Si·bel Oba; Ni·lgün Tanriverdi· International Journal of Psychiatry in Clinical Practice **INDIA**, Volume 7, Number 4, December 2003, pp. 281-283(3)
* **Congenital erythropoietic porphyria in two siblings**; Andréia Jacobo, Hiram Larangeira de Almeida Jr, and Valéria Magalhães Jorge, Dermatology Journal **BRAZIL**; Vol 11, number 5
* **Porphobilmogen deaminase gene mutations in BRAZILIAN acute intermittent porphyria patients**: Ribeiro GS,Marchiori PE, Kuntz Puglia PM, Nagai MA, Dos Santos ML, Nonoyama K, Hirata MH, Barretto OC. Journal of Laboratory Analysis. Pg 259-65
* **Treatment of congenital erythropoietic porphyria in children by allogeneic stem cell transplantation**: P H Shaw, A J Mancini, J P McConnell, D Brown and M Kletzel:, Journal of Association of Physicians of **INDIA**
* **A rare association of acute intermittent porphyria with crohn's** Das, A.K.: Nath, G.L.; Chandrasekar,S.: Dutta.D., Journal of Association of Physicians of **INDIA**

Jennifer Long: Too Young for All of This

The year was 2002 and my life was coming together. I was 25, single, and had a great job where I was loved and respected. I worked since I was 14 years old, so I was finally able to purchase my first home, a little condo that was all mine! A few months later, I began having severe abdominal pain that felt like fire and stabbing. My legs and back also hurt terribly, and I began vomiting but nothing stopped the symptoms. When I improved, I began planning a trip to California, but three days before I was to leave, the pain returned. I went to the same ER four times, until the doctors admitted me and removed my gall bladder. My Uncle Tommy had been diagnosed with porphyria, so we asked for a porphyria test, which was positive for AIP. It was such a relief to finally have a diagnosis, particularly since some medical staff often treated me like a drug addict. (Porphyria patients understand severity of the pain) I was giving up physically, mentally, and spiritually, so I flew to Dr. Herbert Bonkovsky, a porphyria expert, who has dedicated his life to porphyria research. He confirmed the AIP.

Soon thereafter, I became ill again and went to Johns Hopkins where I met with wonderful Dr. Martin. He was not experienced with porphyria, but he wanted to learn. With his care, I improve enough to go on a cruise. The first three days were great, but I became so sick the captain of the ship had the coast guard take me to the closest hospital where I experienced one of my worst attacks and was even hallucinating. My dad had a hard time bringing me home. After spending time in the hospital, I improved. Then I awoke and could not stand. I crawled to my phone and called 911. Within 24 hours, I could not move from the neck down and was terrified. I remained at Hopkins for a month until I was moved for two months of physical therapy. Finally, by God's grace, I am able to walk again. I continue to have attacks and liver problems, but now I can enjoy a life. I believe prayer helped me through all of this.

*Read her full account at the **Patient Stories** section of the APF web site.*

Protect the Future Meeting

The APF and the porphyria experts of the Scientific Advisory Board are deeply concerned about the lack of young academic physicians interested in both basic and clinical research on porphyric disorders. On October 5-6, these experts and the young physicians in training will gather in Houston, Texas to discuss means to protect the future for the many patients who rely on the APF and the porphyria experts for advice and direction for their primary care physicians. The group hope to develop means for supporting Porphyria Centers throughout the country, as well as locating support for the training of future experts.

Your help is needed to support this Protect the Future program. Please mark your donations PTF. Your suggestions for this program are also needed. Contact Desiree at lyonapf@aol.com.

Dr. Kenneth Astrin Departs Mt. Sinai

A friend to porphyria patients, Dr. Kenneth Astrin has departed Mt. Sinai to teach young medical students. He assured us that he will teach them about porphyria.

Dana Doheny, the genetic counselor for the *Protect the Future Program*, will communicate with the patients, as Dr. Astrin's did so kindly and adeptly, while Victoria Njoku will coordinate the porphyria clinic and appointments. Without his help, we could never have completed our new Porphyria Center at Mt. Sinai. Foremost, his kindness to patients who were confused about their diagnosis and was legendary. Desiree said, "Just hearing Dr. Astrin's voice on the other end of the phone made the day happier. People sincerely love Dr. Astrin and will be sad because of his leaving." Thank you Dr. Astrin for your years of support, research and the enormous part you played in developing DNA test and the Porphyria Center for the many people who faced continual misdiagnosis. Don't go too far away!!!!!!!

Dr. Astrin at his desk

Take Note:

Ann, an APF member, had some good suggestions to follow to help educate doctors in her community: *I plan to share my APF information with my primary care doctor and will also share them with all my other doctors at our local hospital. I plan to give the hospital the C.D. and packet of information for their hospitalers and their medical library. Thank you so much.*

The APF will send your doctor information if you will forward his name and address. Remember, however, you can be your doctors best teacher.

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.

What's New on the APF Web Site www.porphyriafoundation.com

Drug Database

Many of you participated in the drug study with Dr. Peter Tishler. The resulting safe and unsafe drug list appears on the web site's home page. However, we need you to help again as the drug list is expanding every day. If you have new drugs information, please contact the APF, and we will forward your input to Dr. Tishler. Also, with Dr. Tishler's help, we are contacting pharmacist organizations to enhance the visibility of our drug database and to advise them that it is a useful tool for dealing with patients with porphyria.

Global Partner

We welcome Japan as our new *Global Partner*. Watch the web site for more information.

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