AMERICAN PORPHYRIA FOUNDATION

1st Quarter, 2014

Simon W. Beaven, MD, PhD The APF welcomes a new Porphyria Physician, Dr. Simon Beavens, who heads the Metabolic Syndrome Program at UCLA. Aside from his experience in metabolic illnesses, he also diagnoses and treats the porphyrias. We are most grateful that such an esteemed physician as Dr. Beavens is available for porphyria patients in the Los Angeles area. Dr. Beaven completed his BS at Stanford University in 1991 (Math with Honors in Humanities), his Premedical Post-Baccalaureate in 1994 at Bennington College, his MD at the University of California, San Francisco in 2001 and his PhD at the University of California, Los Angeles in 2010. He did his Intern and Residency in Medicine at Brigham & Women's Hospital in Boston and his Fellow in Digestive Diseases at UCLA Medical Center, Los Angeles. Dr. Beavens has a host of research publications, abstracts, reviews, book chapters and non-scientific publications. A few of his *Awards, Honors and Academic Appointments include:*

1991	Robert Golden Award for Outstanding Humanities Research, Stanford
1998-1999	Howard Hughes Medical Institute Medical Student Research Fellowship
	Student Research Prize, American Association for the Study of Liver Diseases, 50th Annual Meeting
2001-2003	Clinical Fellow in Medicine, Harvard Medical School
2003-2007	Clinical and Research Fellow in Medicine (Gastroenterology), UCLA
2007-2010	Clinical Instructor and Assistant Professor, Division of Digestive Diseases, UCLA

WHERE THERE IS A WILL-THERE IS A WAY FOR SHAWN WILLIS

My name is Shawn Willis. I am 44 years old and was diagnosed with EPP when I was eight. Prior to my diagnosis, I remember dealing with pain and the long nights with my mom at my bedside doing everything in her power to make me feel more comfortable. Those days were difficult. After my dermatologist correctly diagnosed me with EPP, my life began to improve.

Now I am married to Lori for 23 years and have three non-symptomatic children, Ashley (21), Katie (18) and Austin. Early on in my marriage, Lori and I decided that the sun was not going to keep us from living as normal a life as possible. She has always been a source of strength for me. When she sees a way for me to participate in fun activities, she does whatever it takes to make it happen. Many of you who are active on the APF Facebook page have seen my pictures. I have to give her the credit for the wetsuit idea. At first, I was scared! The thought of being out in the Caribbean ocean or on a remote beach terrified me. But she was convinced that this would work and that I would fully enjoy myself. Well, the rest is history and my wife, once again, was right. She and I took a cruise in February of 2013. We went on a kayak excursion in the open ocean. There I was, kayaking! In full sun! I had the time of my life! It was so wonderful that we decided to take another cruise last year. This time, we took the whole family. We went to Cozumel and Belize. While in Belize, our excursion was to the barrier island pictured here. We had the best vacation ever! The whole family wants to do it again.

I also travel to Uganda once or twice a year for mission work. I don't wear the wet suit in Uganda…ha-ha. That would get a few looks! I rely on my mask and drape hat from Sun Precautions. I haven't yet mentioned the mask and drape hat. Without these items, I would be home bound most of the time. I am cautious not to wear the mask in tricky situations, but most people react well the majority of the time. I wear it in the car, around the house and sometimes in public. I don't go into banks, government buildings or any other place where tensions may be high due to my presence. For instance, while in Cozumel, I took the mask off as I entered retail stores. I didn't go into any jewelry stores, etc. I'm often asked, "Don't you feel weird going into public dressed like that?" My answer is, "YES! Wouldn't you?" But if I don't, then one of two things is going to happen: 1. My children miss out on having an active father in their lives; 2. I end up in pain and still miss out on their lives. It has been very difficult for me to go outside in that get-up, but if I don't, the whole family suffers. This way the entire family wins! So what if I suffer a little bit of a hit on my pride. I have to thank my children and my wife for being strong enough in their character and in their faith to walk in public with me dressed the way I dress. They seem excited to just have me with them. I can't begin to express how that makes me feel as a dad and husband. I will never miss another mission trip, outing or outdoors activity again*!* *Editors note:* ***Thanks for being such a great example Shawn*****.**

WELLS FARGO SUPPORTS THE APF

One of the first members of the APF over thirty years was Janet Molina Veazy. Janet is still a member and continues to support the APF. Six months ago, she called me and we reminisced about old times and how the APF has grown from a handful of members to over 5000 members now. It is such a joy for me to speak with all members, but those who have been friends for such a long time have a special place in my heart. We all struggled together to first, find information on the disease and next, find each other. The APF was the only way. Now the internet lets us find other patients in the touch of the computer keyboard. Janet told me that she had approached the bank where she works, Wells Fargo, and asked them to support the APF with a grant for the *Protect the Future* program to train future porphyria experts. Their answer was a resounding, "YES!" We immediately completed the grant process and were awarded their very sizable grant. Unfortunately, the grant was to be given during the period of Desiree's mother's illness and subsequent death. Being sensitive to her situation and not waiting for her to finally visit the bank, the whole group gathered to send their wishes for great success with the PTF program. They sent the APF this photo of all the bank employees with a copy of the grant check. We sincerely appreciate their generosity and understanding that Desiree was hard pressed to travel. We also want to thank Janet for initiating the process. It is wonderful to see longtime friends still helping the APF plan for the future and care for all those who are newly diagnosed. ***Thank you, Janet and her friends at Wells Fargo!***

A LITTLE BIT OF HISTORY

Porphyria began at the beginning of time and has continued to mutate throughout ensuing generations. The name "porphyria" is only rather recent. For centuries it was known as a blood/liver disease. At one time the abdominal pain was actually thought to be from the liver. The term porphyria itself is derived from the Greek πορφὐρα, *porphyra*, meaning "purple pigment." The name is likely to have been a reference to the purple discoloration of feces and urine when exposed to light in patients during an attack. Although original descriptions are attributed to Hippocrates, the disease was first explained biochemically by Felix Hoppe-Seyler in 1871 and acute porphyrias were described by the Dutch physician Barend Stokvis in 1889. Before, in 1844, Gerardus Johannes Mulder determined the chemical composition of this purplish, iron free substance, which he named "hematin." He also illustrated that hematin took up oxygen. Later, in 1867, J.L.W. Thudichum described the spectrum and fluorescence of these red porphyrins after he published his first book on the analysis of urine.

Based on that, in 1871, Felix Hoppe-Seyler (photo L) crystallized hematin and described it's spectrum. He then demonstrated that the crystalline form differed from one animal species to another. Using his own newly constructed gas pump, he found that oxygen formed a loose, dissociable compound with hemoglobin, which he called "oxyhemoglobin." He renamed the iron free hematin 'hematoPorphyrin.' He is a German physician known for his work in establishing biochemistry as an academic discipline. Felix was the first to obtain lecithin in a pure form and introduced the word proteid (now protein). Additional contributions included metabolic studies and researches on chlorophyll and blood, and especially abovementioned hemoglobin, which he obtained in crystalline form.

In 1874 - Dr. J.H. Schultz first described a case of a 33-year-old male weaver who suffered from skin sensitivity, an enlarged spleen and reddish urine from infancy. He called the condition pempigus leprosus. His was most likely the first description of protoporphyria (EPP). Dr. Schultz was later credited with giving the disease it's name.In 1880 MacMunn described a patient's dark reddish urine during an attack of acute Porphyria. Shortly after in 1888, sulphonal was introduced as a hypnotic drug, Joseph Stokvis (photo L) had a patient, who, after taking the drug, excreated the tell-tale dark reddish urine typical of porphyria. The elderly woman then became paralyzed and died. Stokvis deducted that the pigment in her urine was the hematoporphyrin. Based on that experience, in 1889 B. J. Stokvis published the first case and clinical description of acute hepatic porphyria. CEP porphyria was identified in the year 1923.

In 1930, Hans Fischer (photo R), the Nobel laureate, described heme as the compound that makes blood red and grass green. By 1937 Dr. Waldenstrom in Sweden published his findings. For a time AIP was known as Swedish porphyria, or Waldenstrom's porphyria. In the 1960's porphyria research began in earnest in Europe and in the US. The big break came when scientists were able to recognize ALA and PBG in the 60's. Overall, by the 1960s, all known types of porphyria had been identified and environmental factors were shown to affect the disease course. Research in the 1980s and 1990s led to the identification of the molecular defects in each type of porphyria.

Amanda Boston In late 2011, I started having stomach pains and a lot of nausea and felt sick all the time. When I visited the doctor, he only prescribed Phenergan. My next symptoms were blisters, skin lesions and some big green patches on my chest, hands and arms. When my fingernails fell off, I went to a dermatologist thinking it was some sort of rash, which the doctor treated with every conceivable cream, gel and lotion, but nothing worked. Soon thereafter, I had lunch with friends and passed out. All I remember was a nurse asking me if I knew where I was, but I had no idea. Then I had my first seizure. I knew it was serious when the neurologist tested me and said I had a benign brain tumor. Unfortunately, I began taking a prescription that was unsafe for porphyria. Two days later, I couldn't walk. My parents took me to every hospital in Louisville until they found one willing to take my case seriously. The doctors there said if I hadn't come to that hospital, I would have died that night as my kidneys were failing. They admitted me immediately. My doctor thought I had Guillain Barre Syndrome and started me on plasmapheresis. I was also hallucinating during this time, as well. Three days later I was paralyzed from the nose down and was placed on a ventilator. Doctors were baffled. Fortunately, I had the best nurses ever.

I was paralyzed for three months until a resident doctor thought of Porphyria. I was tested for Porphyria and it came back positive, namely, Variegate Porphyria, which is strange because no one in my family has been diagnosed. I spent eight straight months in the hospital progressing from paralysis to a wheelchair and then a walker. Now I am able to walk. This is not at all where I saw myself at age 26, but I have learned that life is full of unsure times. I am now driving and slowly starting to get my life back. I receive Panhematin every other week to try to control my Porphyria, but I still have attacks at times. Since there is so much pain involved with this disease, I always hope that one day things will get better. I am dedicated to helping with the Panhematin Porphyria research whenever and wherever I am needed, because I really want to help other people. I always say, **"I may not be where I want to be, but I thank God I'm not where I used to be."**

NEW ON APF TEAM I would like to introduce myself to You. My name is Natalia Sturza, and I am the new Director of Development at the American Porphyria Foundation. I was born in Western Europe, in a small country called Moldova. When I was 11 years old, my family and I moved to Russia where I finished high school and graduated from the State University with a Master Degree in Mass Communications. During my University years, I participated in students' exchange programs and was, therefore, able to travel to the USA for the summertime. I always found a joy in meeting new people and learning new cultures. When I moved to the USA several years ago, I attended Texas A&M University – Central Texas and earned an MBA degree. In my spare time, I enjoy active types of sports – mountain biking, skateboarding and snowboarding, which is quite a difficult task for Texas ☺

I am so honored to be the Director of Development at the APF. I am thankful for the opportunity to be a part of a great Team, to contribute to the porphyria community and be able to make a difference. I will be working on many different projects to heighten awareness of porphyria and will be collaborating on research with the research coordinators at the six porphyria research centers. I hope my efforts will help more people to be diagnosed promptly and accurately and receive proper treatment. *Natalia*

PORPHYRIA DOCTOR FINDER Every day the APF receives requests for a doctor who can diagnose and treat porphyria. The main question we hear is, "Where can I find a doctor to treat me?" In the past, the APF has only given out the names of doctors who were experts in the porphyrias, but the number of experts is very small. However, our policy has changed a bit since there are now 5000 members and so few experts. Many of our members have told us that their doctor has some measure of knowledge about the porphyrias and that their doctors diagnosed and treated them well. A few years ago, we began to collect the names of those doctors around the country and placed them on our database. We also sent out several thousand very comprehensive Physician Education packets to doctors who were interested in learning about porphyria. At the behest of many of you, we created FIND A DOCTOR section on the APF website. Check the FIND A DOCTOR section and see if a city near you is listed. Then call the APF for the doctor's contact information. Watch the website often as the section will be updated often. Experts are also listed for an appointment. If you have a great doctor, please contact the APF. Note we cannot validate the level of knowledge of any of the listed doctors other than the porphyria experts. ***See www.porphyriafoundation.com.***

WHY WAS MAD KING GEORGE MAD Last year a remarkable exhibit came to light. Hidden in the vaults of a London museum was a scrap of paper containing a few strands of hair. The paper was crudely fashioned into an envelope but the words on it immediately caused a stir: "Hair of His Late Majesty, King George III." For Professor Martin Warren, it was the clue that would help him finally solve the mystery of King George's illness. His investigation is featured in a BBC documentary, *Medical Mysteries*. "King George is largely remembered for those periods when he lost his mind. But it's been difficult to explain these attacks, so I was keen to analyze this hair sample," said Professor Warren. When the hair was tested by the Harwell International Business Centre for Science & Technology, the results were surprising. The king's hair contained over 300 times the toxic level of arsenic.


King George's wigs full of arsenic

Far from being an answer, this remarkable finding was just the start of Warren's detective work. In King George's time, his bizarre behavior and wild outbursts were treated as insanity. He was bound in a straitjacket and chained to a chair to control his ravings. King George was officially mad. It wasn't until the 1970s that a new and controversial diagnosis was made by two psychiatrists, Ida MacAlpine and her son, Richard Hunter, who revisited the king's medical records and noticed a key symptom; dark red urine, which is a classic indicator of porphyria. They also noted the severity of the King's attacks and that his attacks were after age 50.

Professor Warren (photo L) knew that porphyria attacks can be triggered by a wide range of substances–alcohol, certain medications, even monthly hormones. Perhaps arsenic could also be a trigger, so he contacted Professor Tim Cox, an expert on extreme cases of porphyria at Addenbrookes Hospital in Cambridge. Professor Cox confirmed his guess– arsenic was listed as a trigger. And the massive levels found in King George's hair suggested that the arsenic had been liberally ingested over a long period of time. According to Warren, the two professors began poring over the King's medical records preserved in the Royal archive at Windsor. There was passing reference to arsenic used as a skin cream and as wig powder but nothing that could explain the staggering levels of arsenic showing up in the king's hair. The most common medication he was given was James' powders, a routine medicine he was being given several times a day–made of a substance called antimony. Tracking down James' powders at the Royal Pharmaceutical Society, Warren found the final piece of the puzzle in a 19th century almanac. Antimony, even when purified, contains significant traces of arsenic. Warren believes that the arsenic from the very medication King George was being given to control his "madness" was triggering more attacks. Warren feels that the porphyric attacks had been brought on after a lifetime of arsenic accumulated in his body and were made much more prolonged and more severe by the medicine to treat him. Warren said, "It is a very convincing explanation of the king's attacks, and could account for why he had them at such a late stage in life and why they were so severe. *Editor's note: Many experts disagree with Hunter about the King having an acute porphyria. However, a friend of mine is a British Lord whose wife is in the lineage of King George III. Her brother had an acute porphyria and suffered many acute attacks before he died. Although this is significant evidence, it is not definitive for the question.*

PAIN, PAIN GO AWAY For almost a year now, the APF has been instituting a major program to change the new pain laws that keep patients with porphyria from being treated properly for pain. Porphyria patients are the unintended victims of such legislation. For people who are in terrible pain, like porphyria patients, this has become a huge problem, especially since not all pain medications are safe for acute porphyria patients. In certain states, a patient's primary care physician cannot prescribe special levels of drugs. Patients are sent to pain clinics where doctors generally know little to nothing about porphyria. Plus, pain clinics often require a $500 entry fee and a wait of 1-6 weeks for the first appointment. For a person in horrible pain, this is a horrible problem. **Dr. Lisa Kehrberg,** who is a pain management physician, was recently diagnosed with acute intermittent porphyria (AIP). Because Dr. Kehrberg endured extremely severe pain during her many acute attacks, she personally understands the problem of porphyria pain. In fact, she said the pain was unbearable, unimaginable and she felt great empathy for other patients enduring attacks. As a pain management physician, she felt she was a very understanding and helpful doctor, however, now she has an even greater sensitivity to her patients.

Fortunately, in March Dr. Kehrberg will be filming a DVD that the APF will use for our physician education program to help doctors also understand the unbearable pain that occurs with porphyria attacks and the nerve damage that can occur after the attack, as well as the need for appropriate pain treatment. We hope to have the DVD on the APF website in late March. Dr. Kehrberg will also be assisting Desiree Lyon Howe, APF Executive Director, with our APF Pain Programs in specific states, like Florida, where pain laws are harmful to people in pain.

NATIONAL PORPHYRIA AWARNESS WEEK, April 20-26, 2014

The challenge of living with porphyria starts with how little is known about it among friends, family and the medical community. Although the APF staff uses many means to promote porphyria awareness, YOU are the most effective means, because YOU are in the doctor's offices, Emergency Rooms and around your community. During **National Porphyria Awareness Week (NPAW)**, April 20-26, 2014, YOU have the opportunity to enhance porphyria awareness in your hometown. For example, the Cook family got their hometown involved by creating Hat Day, to heighten awareness of EPP. Their sons, Caul and Cason, who have EPP, explain their severe photosensitivity and how the sunlight causes them severe pain and swelling. The schools allowed students to wear a Hat and bring a dollar for the APF. Over a few years, the event has grown to reach churches, clinics, hospitals, TV and news media, etc. *Polly Havard ran a marathon, Andrew Turell interested a group in research, Rob Saupé walked 100 miles, Lisa Kancsar and Diana Ijames set up porphyria exhibits, Michael Drew and Charles Johnson manned booths at medical conventions, the Fleegels filmed an EPP documentary. Carrie Hunter created a fundraising page and 100 others helped with awareness event.*

Below are suggestions for you to create your own awareness event. The APF will provide: Porphyria Brochures, a *Porphyria Live* DVD, Fact Sheets, Powerpoint Presentations and educational materials for physicians and hospitals and press releases for local newspapers and television and other suggestions.

SHARE knowledge about porphyria in your doctor's offices and local hospitals. Suggest that they host a seminar or grand rounds on porphyria. Ask if there is a local meeting where you can hand out materials.
ASSIST at medical conventions or health fairs to educate laypersons and physicians on porphyria.
TELL your story to local media. Television, newspapers, community magazines are looking for human interest stories about people who have encountered a major illness and have undertaken the challenge.
VOLUNTEER your talents or skills to help achieve the educational programs of the APF. For example, donate your paintings, sculpture, computer expertise, business acumen, etc. for our fall raffle or to help APF.
HOLD a community race, car wash or other fund raising activity.
HONOR your loved one with a gift to the APF for a birthday, anniversary, holiday or memorial tribute.
WRITE a letter to friends and family asking them to consider making a donation to the APF.
ASK your local newspaper or community newsletter to include a story about you and porphyria or just write about your porphyria for your family or the APF website or newsletter.
LEARN how to be an advocate in your daily life. Opportunities to share your knowledge are everywhere.
BEFRIEND your physicians, and they will share their new found knowledge of the disease.
HOST a patient meeting and share your experiences and your successes in advancing porphyria awareness.

PATIENT MEETING

The APF hosted a terrific patient education meeting at the Marriott Hotel in Bethesda, MD. The gathering was unusual in that this is the first time we have had testing during the meeting. Over 50 people attended the meeting to hear porphyria expert, Dr. Karl Anderson, make a presentation on the porphyrias and answer questions from the attendees for almost three hours. Also, there was Dr. Robert Desnick, who oversaw DNA testing of those who wanted to join the research studies and discussed the research with patients as shown in the below photo. Clinical research coordinator, Ms. Hetanshi Naik, helped the patients register for the trial and perform the cheek swabs for the DNA. We were also fortunate that renowned porphyria researcher and Consortium member, Dr. John Phillips, from the University of Utah, attended and helped answer individual's questions after the meeting. One of the interesting aspects of the meeting is that we had a number of families who attended and enrolled in the research. Ariel Lager, an APF member who came from New York, shared her experience as one of the eight research patients in the genetic studies in Spain, as well as the US research projects.

The meeting was a huge success and everyone agreed that they were headed home with far more knowledge than when they arrived. The APF will host more meetings in settings surrounding Consortium meetings and medical conventions like, ***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. We hope to also host another meeting in NYC in May and others throughout the year. For information on the next meetings, watch YOUR weekly ENEWS. In you are not receiving the APF ENEWS, please call the APF and give them your email address or your new email address. Sometimes people forget to update us when they have an email or address change. Thank you, Hetanshi and Drs. Anderson, Desnick and Phillips for your help.

PICC vs PORT: *My name is Benjamin Stratz. In 2010, I was diagnosed with Variegate Porphyria and am a third generation VP sufferer. I am grateful to my doctors, the APF and all the people in my life who have been supportive. I hope this synopsis of my experience with PICC's and a Port is helpful.*

Many people diagnosed with an acute porphyria are not familiar with how the Panhematin treatment is administered and what is expected during treatment. The manufacturer, Recordati Rare Diseases, recommends that Panhematin be administered through a central line to prevent the possibility of phlebitis. However, your doctor may recommend that you have either a Peripherally Inserted Catheter (PICC Line) or a Central Venous Catheter (commonly called a "port"). My experience follows, but your decision should be discussed with your Doctor.

The PICC Line generally looks like a regular IV except it accesses your circulatory system from a peripheral line near the surface and is surgically placed in a vein inside your body (usually one of your limbs) where it is manually fed toward your heart and extends outside the body about 4-6 inches. Blood can be drawn from this line and medications can be administered. I have had four PICC lines, two in my right arm (just below the bicep, one in my left arm and one in my chest. This procedure lasted about 30 minutes under local anesthesia. I had no pain during this procedure, but the site was a little sore for a few days afterwards. When I first started receiving Panhematin via the PICC, I had few limitations as long as the line was properly wrapped and kept clean and dry. I was cautious not to get it caught on anything. Bathing was probably the most challenging aspect of having a PICC, so unfortunately, swimming is not an option. During my Panhematin treatments, I also received daily IV fluids at the hospital. When my treatment was complete, the doctors removed the PICC line, which only lasted a few minutes and was painless. I have a small, 1/16" scar at the site of each PICC line. After being hospitalized several times and receiving a new PICC each time, the doctors recommended that I consider having a Port.

The Port procedure is more involved than a PICC line. In June 2013, the Port was installed under general anesthesia. I was in a considerable amount of pain after the procedure. Both the site of the port and my left shoulder/arm were painful for several days afterwards. It took about two weeks for the pain and shoulder stiffness to subside. The Port was placed on the left side of my chest a few inches below my collar bone. All that is visible is a half dollar coin sized round bump. Like a PICC line, a tube goes from the access point (the visible bump) to a central vein close to the heart. After the surgery, an X-ray verified the port position and the surgeon Ok'ed its use, which for me was within 24 hours. Since the port was under the skin, I didn't have to maintain the lines and could bathe and swim. I was told that the port could last up to 10 years with proper maintenance. Aside from the initial pain, of the port, I am much happier with it opposed to the PICC line and haven't had major problems with it. The port site is sometimes painful but when compared to the suffering associated with an attack, the discomfort is worth it.

Editor's note: Many different complications can and do occur with both the PICC and PORT.

POLLY RACES FOR EPP **Polly Barton Havard** did not let any grass grow under her feet when she heard that the APF was already preparing for Porphyria Awareness Week. She planned her awareness event around a New Orleans race. Polly completed the four hour, 26.2 mile Rock and Roll Marathon in New Orleans, Louisiana on February 2nd. Her goal was to use the event and her participation to heighten awareness of Erythropoeitic Protoporphyria (EPP), because her daughter, Madelynn suffers from the disease. Polly also raised funds for the APF through her online donation Firstgiving Page. Her goal was to raise $1500, but she almost doubled her goal by raising $2600. ***Thanks from all of us, Polly !!!***

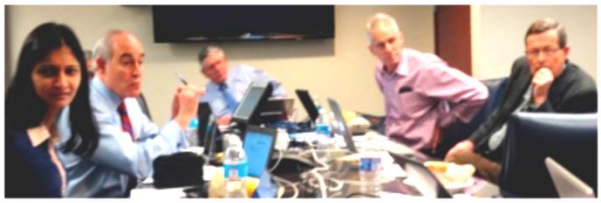
RAPID PBG TEST The Rapid PBG (porphobilinogen) test is a random, rapid, "screening," procedure for excluding an acute porphyria attack or identifying those patients who need more specialized investigation all within 20 minutes, rather than the usual 2 weeks. The absence of increased urinary PBG in a suspected attack excludes the diagnosis. Some patients, especially with acute intermittent porphyria, excrete excess PBG even in remission, but in an attack, this increases above their general concentrations. Thus, "screening" of urine for excess PBG and its measurement have an essential role in the initial diagnosis and management of such patients and in their continuing care. A positive result should be referred to a Porphyria Reference Laboratory where the blood, urine and stool can be studied. In this way the diagnosis of acute porphyria can be confirmed and family studies carried out. Read about the Rapid PBG Test:
http://www.porphyriafoundation.com/sites/default/files/tiny_mce/filemanager/files/testing_treatment/PBG.pdf.

Thermo Scientific is considering abandoning the production of this Rapid PBG Test because so few are being used. This would be tragic for porphyria patients. Please encourage your hospitals to order this 20 minute diagnostic test.

RESEARCH CONSORTIUM MEETS

We are all very fortunate because the researchers who are members of the Porphyria Research Consortium meets quarterly to discuss their research and their most recent discoveries. Recently, the Consortium met in Bethesda, MD, which means they met to find means to help YOU via their research. They need YOU to participate in these studies.

Photo: H. Naik, Drs. Desnick, Bonkovsky, Bissell, Anderson



People with the following are needed:

- have recurrent acute attacks
- have X-Linked EPP,
- have recurrent attacks for a Panhematin study,
- have PCT for a Phlebotomy vs Hydrochloriquine.
- have all types for a Longitudinal Study

Included in their agenda was an update on the Longitudinal Study and a discussion of their present research:
Dr. Joseph Bloomer, University of Alabama, Birmingham, AL-Mitoferrin 1 Expression in EPP
Dr. Karl Anderson, University of Texas, Medical Branch, Galveston, TX- Panhematin for Acute Porphyrias
Dr. Montgomery Bissell, University of California, San Francisco, CA-Clinical Diagnosis of Acute Porphyrias
Dr. John Phillips, University of Utah, Salt Lake City, UT-INH for EPP
Dr. Karl Anderson, University of Texas, Medical Branch, Galveston, TX-Phlebotomy vs HCQ for PCT
Ms. Hetanshi Naik, Mount Sinai School of Medicine, New York City, NY-EPP Natural History
Dr. Robert Desnick, Mount Sinai School of Medicine, New York City, NY-Capture Array for DNA Studies Update
Dr. Karl Anderson, University of Texas, Medical Branch, Galveston, TX-Four Informative Acute Porphyria Cases
Dr. Herbert Bonkovsky, Carolinas Healthcare System, Charlotte, NC-Oral Iron Absorption in EPP patients in Controls
All Consortium - Natural History of Recurrent Patients and High Excreters, plus the upcoming Alnylam research.

REMEMBER, RESEARCH IS THE KEY TO YOUR CURE!

GLUCOSE EFFECT

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 acute 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 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 patients who don't understand that this simple treatment involves a very complex mechanism. **Please note that guidelines suggest that when a person is <u>having symptoms</u> of porphyria, the carbohydrate intake be increased.** It is not necessary nor is it healthy to become overweight due to carbohydrate loading when symptomatic. *See Diet Section on our website.*

VOLUNTEER AT A MEDICAL CONVENTION

You 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 who will diagnose people with the acute porphyrias. Your help is needed to man the booth, so please call the APF to volunteer and receive more information about the conventions and your time there.

What's New at the APF

www.porphyriafoundation.com

Is Your Membership and Contact Info Up to Date? The APF is able to maintain our physician and patient education programs and many other services because of your support. Since we do not receive government funding, we need your support and donations. We also need your new contact information if you have a new address or email. Be sure to send us your email address so you can receive the ENEWS.

Protect the Future program to train future experts is important. Please consider making a donation to this program. Yours and your children's future health depends on each of us supporting the training of doctors who will know how to treat us and perform research when our present experts retire. We have the opportunity to fund this training now so that the present expertise is not lost. Please send your donations to the APF and mark them PTF. They will be placed in the PTF fund to be used for the training of young doctors as future porphyria experts.