AMERICAN PORPHYRIA FOUNDATION

1st Quarter, 2007

Lauren Warren: A Porphyric Pollyanna

My life began again this past December. You see, I left the physical healthy person I was at twelve years old when my symptoms became so severe that I was rushed to Emergency Room where I received the first of many misdiagnoses. Fortunately, the disease was sporadic enabling me to live a normal life between the terrible bouts of the mystery disease. It has been a long journey; one I am sorry to say is all too familiar with most of the readers of this story. Two months ago, I had a bad attack of a mystery illness that has plagued me for 28 years. On that night, I asked to go to hospital because of Indescribable pain, fear, and weakness. My medical team had nothing to offer but a phone number of a hematologist. Because I was desperate for an answer, my partner gave me the saved birthday money for a trip to the doctor. The gift was to be used to take me on vacation to celebrate my 40th birthday, but I had a better present. I made the two-hour drive to Dr. Marshall's office where I received the best gift ever: HOPE. Within five minutes, Dr Marshall, felt he had the answer, which tests later bore out as acute porphyria. I finally have an answer to my unrelenting pain and weakness. I believe I serve an awesome God and am grateful for His mercy.

To help the APF's National Porphyria Awareness Week, I signed up to participate in the **Lake Placid Ironman Race** in July, Being part of the **"Janus Challenge"** is important in that an athlete can name any foundation of their choice, raise funds for that foundation, and the **"Janus Group"** will match a portion of what is raised. I am excited to be alive, to have such incredible hope, to know how to hopefully decrease the frequency and severity of the attacks. More importantly I am thrilled to help others by raising money for the **American Porphyria Foundation**. I have often been teased that I have a Pollyanna attitude toward life, even during the tough times. I guess nothing has changed except now I am a Porphyric Pollyanna and, on July 22nd 2007, I will cross that **Lake Placid Ironman Finish Line** for us all, and I will carry you in my heart and thoughts every step of the way and for me there is only one direction until I reach the finish line.

For more of Lauren's story, read **www.porphyriafoundation.com and click on "Patient Stories."** *Editors note:* ***To help Lauren in her effort to raise funds and porphyria awareness through the Janus Challenge for the Lake Placid Ironman Race, send your gift to the APF and mark it Janus.***

IN TOUCH MEETS

On Saturday, January 13th, 2007, Sarah and Andrey Korobovsky hosted an IN TOUCH meeting at the Moreau Emergency Squad in South Glens Falls, NY. Sarah, who has AIP, wanted to network with others with porphyria and got the idea after attending the IN TOUCH meeting in Yonkers, NY, in October. "I met with other people, who like me, had their frustrations and successes, and wanted to provide a similar forum here in Upstate New York. My husband, Andrey, and I traveled about 3 hours in October to attend the IN TOUCH, hosted by Patsy Brady. Patsy was lucky enough that Desiree and Dr. Poh were in the area to have them present and answer many questions, I was hoping to do something positive as well. Half a dozen people came from two to four hours drive away. Working closely with Lelia Brougher at the APF, we were able to teleconference with Dr. Herbert Bonkovsky, a porphyria specialist in Connecticut. As a service of the APF, Dr. Bonkovsky spent over an hour explaining porphyria and answering questions. He also provided detailed points of contact with phone numbers and hospital affiliations for the Upstate New York region, Vermont and Massachusetts. All the attendees either have AIP, or were significant others of people with AIP, which made the event that much more relevant as there were many common threads running through the stories exchanged.

After the call, we viewed the APF material and talked about our porphyria journeys. There were many things that we all shared in common from the 'what's happening to me' phase to finally being diagnosed and being able to understand what's going on in our bodies. After the meeting everyone went to a local restaurant and talked for another couple of hours over dinner. It was fulfilling to make these contacts and bring credible information to others with AIP. Maybe we'll do this again next year and reach out to more people."

JOIN THE IN TOUCH NETWORK AND RECEIVE THE NAMES OF ALL OTHER IN TOUCH MEMBERS.

CONTACT THE APF TOLL FREE: 1.866.273.3635

Connecticut Protect the Future

We welcome **Dr. Manish Thapar** (left) as one of our *Dr. Cecil Watson, PROTECT THE FUTURE* candidates. Our PROTECT THE FUTURE program was established to interest and train brilliant young physicians as our future porphyria experts. Dr. Thapar is a Fellow in Clinical Research, Advanced Hepatology and Liver Transplantation at the University of Connecticut Health Center in Framington. His training with renowned porphyria expert, Dr. Herbert Bonkovsky, (right) was made possible by a generous APF family.

Dr. Tharpar has had a long-standing interest in Hepatology and metabolic disorders dating back to his days as a medical student in India. Guided by his quest for professional satisfaction, Dr. Tharpar joined an internal medicine residency program in the United States after completing his medical training in India. His experience here has been in his words "intellectually and academically fulfilling." Dr Tharpar resides in Framington with his wife, Deepi, also a physician, and his three year old daughter, Mehek. Together he and Dr. Bonkoksky have already completed several major porphyria publications below:

- Bonkovsky HL and Thapar M; Porphyrias, Conn's Current Therapy by Rakel, R.E. and Bope, E.T., Elsevier health, 60th Edition, 2008 – in press.
- Lambrecht RW, Thapar M and Bonkovsky HL; The genetic and genomic aspects of Porphyria Cutnea Tarda, Seminars in Liver Disease, 2007 – in press.
- Thapar M and Bonkovsky HL; Variegate Porphyria, Encyclopedia of Molecular Mechanisms of Disease, Springer-Verlag, 2007 – in preparation.

Editor's note: *Recently, I was asked what I felt was the most important need we should undertake at the APF other than a cure, which is, of course, our greatest need. I immediately told them at our* ***Protect The Future*** *program was of utmost importance, because over the next decade, we will lose ninety percent of our valued Porphyria experts to retirement. These men and women have led the field of Porphyria research, testing and treatment for the past thirty years. Now there are less than a dozen porphyria experts in the US. With their retirement, we run the risk of losing knowledge of the disease, quality testing, diagnosis, treatment and ultimately a cure. Together we can change this situation by supporting the* ***Protect The Future Program To Train Porphyria Experts For Our Future****. Mark your donation* ***PTF****.*

Rotterdam Porphyrins & Porphyrias Meeting

Researchers and Clinicians are invited to attend the 2007 Porphyrins and Porphyrias Meeting in Rotterdam, The Netherlands, from April 29th till May 3rd in 2007. These biennial events have been held around the world for many decades. The meeting will begin with a welcome party on Sunday evening April 29th and close midday May 3rd. Also, there will be a special clinical session on May 1 from 10:30 hrs – 16:30 hrs to discuss specific patient cases and hear a patient presentation.

The presentations and discussions will cover subjects such as, heme synthesis, enzymes, structure and function, molecular/genetic aspects and clinical syndromes, heme degradation (including heme oxygenase, bilirubin aemias, etc.) iron related topics, general porphyria diagnostics, therapies and clinical issues, and drug porphyrinogenicity.

The aim of the meeting is to create an environment for active participation for experienced and new researchers in the field, in an informal atmosphere with lectures and poster sessions. The APF continues to support these important meetings and to assist in encouraging new experts in the field. We hope that all porphyria experts and researchers will attend.

JOIN THE APF ENEWS . . . CONTACT THE APF TOLL FREE: 1.866.273.3635

PBG Screening Tests for Acute Porphyria

The APF has made a major impact in the use of **ThermoFisher Scientific PBG** (porphobilinogen) **Screening Tests**, because they are excellent for quick screening for the acute porphyrias. Prompt results are extremely important so that the patient does not suffer with an attack while awaiting lengthy tests. These tests are relatively painless and are performed using a small 1 ml random urine sample without preservatives instead of a 24-hour collection and within 20 minutes the results are returned. Also, testing can be performed within 8 hrs of the collection or stored at 20°C. The **PBG Screening Tests** are manufactured by ThermoFisher Scientific, Inc. in Pittsburgh, PA, 1-800-558-9115. Unfortunately, not many laboratories have these PBG testing kits. However, the APF has begun to make a measurable impact on the use of these kits. With your help, we can make an even greater impact. Every emergency room and reference lab should have access to this kit. By informing our doctors, their reference laboratories and ERs about the **ThermoFisher PBG Screening Test Kit** for Acute Attacks, we can help others receive a porphyria quick diagnosis. Remember, these tests require laboratory skill and are **not for home use**.

How it works is: During an acute attack of Porphyria, the heme precursor, PBG, accumulates in the liver. Raised levels occur in the plasma and urine. An increase in urinary PBG, therefore, is strongly indicative of an acute Porphyria attack. The Trace PBG screening method utilizes an anion exchange resin, which binds PBG present in the urine. Interfering compounds are then removed through a simple washing step. PBG is then eluted off the resin and added to a DMAB/acid solution. If PBG is present in the sample in abnormal amounts, a magenta color develops. By comparing the color developed with the standards included in the kit, an approximate concentration of PBG can be determined.

While quantitative tests require a high level of expertise, The PBG Screening Kit can be used in most reference laboratories. See: www.thermofisher.com. If the confirmation assay confirms acute Porphyria, it is strongly recommended that further investigation to define the type of Porphyria be performed at a laboratory specializing in the porphyrias. The following are a few labs that have the tests kits. Check the APF website for other labs and ask for your lab and Emergency Room to use them, as well.

South Bend Medical Found	South Bend	IN
Memorial Med Center	Jacksonville	FL
Univ Hospitals of Cleveland	Cleveland	OH
Parkland Health and Hospital	Dallas	TX
Mission Hospital	Asheville	NC
Univ of Rochester Hospital	Rochester	NY
Vanderbilt Univ Hospital	Nashville	TN
Psychiatric Associates	Parkville	MO
Kernode Clinic	Burlington	NC
Univ of Alabama Hospital	Birmingham	AL
Sarasota Memorial Hospital	Sarasota	FL
St Joseph's Womens Hospital	Atlanta	GA
Brigham and Women's Hosp.	Boston	MA

"My wife just had a terrible acute attack of porphyria, which was due to a late diagnosis of the disorder. The doctors didn't suspect porphyria until they realized she was only getting worse after a four-week hospitalization, receiving intensive care treatment for misdiagnosed Guilan-Barré syndrome. Although still restricted to a wheelchair,she is improving while being treated by a physiotherapist to regain full control of her body. Having experienced the difficulties of a late diagnosis of this terrible disease, I would like to help others to avoid such a traumatic experience. All my wife's relatives are at-risk individuals, including my 17 year-old daughter. I am sure that the early diagnosis provided by this test kit will help, not only our relatives, but a much larger number of folks with porphyria throughout the country." Douglas Bassoli

If you would like your physician, laboratory or hospital to receive information about this test kit, please contact the APF and give us their names and contact data.

Laboratories in Europe

We have been asked often how the laboratory and testing system for porphyria differs in Europe from the United States. Interestingly, this and many testing comparisons were addressed at an international meeting several years ago from which we were able to glean their summarizations. The following is a presentation entitled, *A View From America* by Dr. Karl Anderson, porphyria expert at the University of Texas Medical Branch in Galveston, Texas.

Most European countries have one or more specialized university-based laboratories that are resources for the biochemical and molecular diagnosis of porphyrias. These laboratories and associated clinical and laboratory scientists are funded largely by national governments research and service, and they provide expertise for reliable diagnosis and specialized treatment of porphyrias throughout Europe. In some European countries, most families with inherited porphyrias appear to have been identified and their mutations are known. As a result, it seems possible most gene carriers in such countries can be identified, and preventive measures taken to avoid symptoms of porphyria.

By contrast, in the US, where federal government funding for porphyria research has declined, there is only one university laboratory that is a resource for biochemical testing, and another that is beginning to offer DNA testing. Most porphyria testing in the US is performed by large commercial laboratories that report results that are of uncertain reliability and are often mis-interpreted. Therefore, there is often substantial delay in diagnosis, the majority of patients and their families are newly recognized, and it is likely that there are many unrecognized individuals and families with porphyria.

This is why it is very important for you, our members, to become acquainted with the information in the APF website so that you can be sure you are being given the correct test and can insist that the test is handled properly.
See: www.porphyriafoundation.com

Charles Johnson's Good Solution For Serious Problem

Charles Johnson of Brighton, Massachusetts was surprised when he awakened with "gut wrenching, unimaginable pain" one Saturday morning nine years ago. He rushed to the ER where he was treated for dehydration and sent home. He told the doctors that his mother had died of AIP, but the revealing information was ignored.

After this cycle occurred a few times, he went to Mass General Hospital for help. Fortunately, a young physician tested him for AIP and discovered that he had AIP. However, the attacks continued to occur so often that he was hospitalized every six weeks. Last year, Charles was hospitalized for six months. He was prescribed Panhematin infusions and they stopped the attack. Unfortunately, they would occur again in a few weeks.

Desperate to control the attacks, Charles wanted to try Panhematin infusions on a maintenance basis every two weeks to stabilize his ongoing attacks. He discussed his idea with porphyria expert, Dr. Herbert Bonkovsky, who thought it was a good one. But his own doctors at St. Elizabeth's in Brighton had never tried the bimonthly Panhematin regimen and were reluctant that it would work. Charles approached them and used Desiree's "thousand flaming swords" pain analogy to encourage them to try it. They quickly agreed.

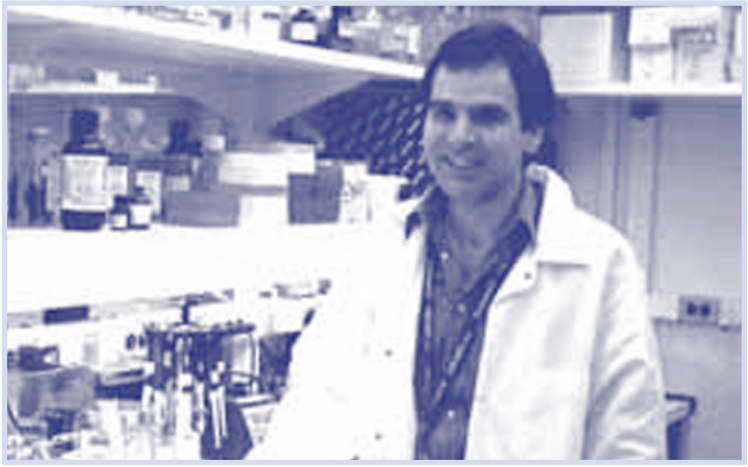
The bimonthly regimen has been used well with many other people, but it was new to Charles and his doctors. Charles began the Panhematin infusions every two weeks at the chemo infusion section of the hospital, and his life changed dramatically. He was hospitalized only three times last year, instead the usual hospitalization every few weeks. Since being his new Panhematin regimen, Charles' general health and strength have improved so much that he can now return to work. He also credits his wife, Judy, who never lost hope and cared for him with a smile…all this while taking care of four children, as well. Charles does not want others to suffer as he has, so he joined and will continue the APF efforts for the National Porphyria Awareness Week.

Of his experience, Charles says, "I still have a good life. There are so many caring people in the world; my family, friends and neighbors have been so helpful and understanding. My wife, Judy, is my rock. She is always smiling, loving, caring. Without her, I could not have made it. I have missed many holidays and sporting events for my kids, and I cannot predict when I can go somewhere. But my mind is always busy, my phone is always ringing and I am blessed to be healthy in every other way but AIP.

National Porphyria Awareness Week

The NPAW, February 17-24, was the kick-off for our members to help heighten awareness of the porphyrias. Many of our members helped with this most important mission. Some contacted their local media, asked their regional hospitals to do in-service programs and seminars, and a variety of other awareness activities.

It is not too late to participate. By spreading information about porphyria, the public and physicians alike will more readily recognize the need for better diagnosis and treatment.



In Memory

Dr. Eric Lemmer We are saddened by the recent passing of Dr. Eric Lemmer and join the NYC Mt.Sinai Hospital in expressing our sympathy to his family. Dr. Lemmer was a warm, compassionate clinician, as well as an inspiring, beloved teacher and researcher. Dr. Lemmer will be missed by all who were privileged to know him.

Sadly, Dr. Lemmer was to become one of the Protect the Future trainees and had studied porphyria at the famous Cape Town Porphyria Center. He was one of America's most outstanding young gastroenterologists and was selected as a 2006 American Gastroenterology Association Research Scholars.

In Honor

These individuals were honored by loved ones with a generous gift to the APF.

Dr. William Weaver **from Dr. Peter Tishler;** ***Judy Roberts Lucero*** **from Beverly and Larry Roberts;** ***Dr. Lee Smith*** **and** ***Geri, Dr. William Sawchuk*** **from Bonnie Marcus;** ***John Saulmon*** **from Nancy Saulmon;** ***Dan and Aaron Pudlicki*** **from Sandy Pudlicki;** ***Alice Patricia Brady*** **from Pamela J Marin;** ***Audrey and Ken Timper*** **from Diane Paquet;** ***Dr. Karl Anderson, Desiree Lyon*** **from Judy and Tom Phelps;** ***Ralph M. Gray*** **from Gary Eyster**

What Doesn't Kill You Makes You Stronger

Sixteen year old, Craig Leppert, says, "What Doesn't Kill You Makes You Stronger." It is the motto of the Leppert family. They should know! Craig and his sister, Nicole, have Erythropoietic Protoporphyria EPP and are champions at taking on the many challenges they face to live with sunlight as their enemy. According to Craig, he showed signs of EPP at 18 months old. So as to not appear different from his peers when he was in grade school, Craig stayed in the sun to play even though he knew he would suffer the consequences later and suffer he did. At times he was plagued with severe burning that would continue all night. Now he tries to protect himself from the sun's harmful effect. This is, of course, hard to do when you are your high-school's starting linebacker. Nonetheless, Craig puts on his gloves, tinted visor, long turtle neck, and "Under Armour" sun-protective sports apparel for football practice and games. Moreover, he has a good attitude, which he developed during the long days and nights of severe burning. "You learn a lot about yourself and your faith at 3 AM when you are dealing with severe pain," says Craig. "You even learn to live past, 'Why me, God' and learn that instead He has given me a special opportunity to become a stronger and better person." Along with developing his inner strength to help control the disease, he also takes Lumitene for the EPP. Even still, he often has to bath in ice packs to alleviate the burning. But one day Craig will have his dream to go to the beach, because he says, **"I believe in EPP expert, Dr. Michelene Mathews-Roth, and I know that she will find a cure. It is only a matter of time."**

NICOLE: "My name is Nicole Leppert. I am 12 years old, and I have EPP as does my older brother, Craig! I really don't remember having symptoms for the first time, but I can remember in Preschool and Kindergarten having to wear protective clothing and hats for recess. I don't have a lot of symptoms, because I don't like to put myself into the sun. I know how it feels, and I don't want to have to go through the pain! When I do get sun exposure, it hurts, and when I say it hurts I mean IT HURTS!!! There's no really good way that I can explain the pain, but by just saying it itches, burns, and feels like your on fire! Most times you feel fine, then it'll just strike you! Mainly, it burns you from the inside out! But I don't sit just inside all day; I joined the Student Council and I'm taking dance this year. I go to the movies, and have a lot of friends, who treat me like a normal person with a sun disorder. Sometimes I feel, "Why me?" I can't blame anybody for this, not my parents, not my family, not God, not myself–It just happened. We can live past EPP and understand that we are special and different! I think it's harder for boys with EPP, because they play sports and girls like the MALL. I love to shop till I drop!

This disorder can help you find out what kind of person you are; what you can handle. It helps to have my whole family and friends as a huge support system. It helped me to see Craig walk out at his 8th grade graduation two years ago, all covered up from head to toe with his head held high! "I knew then I could accomplish anything!" *The Leppert Family and Craig's High School are raising money to purchase special lights to enable Craig to play football under the night sky. If you would lke to help, please contact the APF.*

Family and friends have made donations to the APF in memory of their loved ones. Lorraine Dunlop lost her husband, ***Donald Dunlop***, who had AIP. These friends and family remembered him with a gift to the APF:

Butch and Jean Kasprowicz, Maxine Larson, Bea and Ken Johnson, Bonnie and Gilbert Sampel, Joyce and Dick Paschke, Marge and Bernie Peterson, Alice Sexton, Roberta Jensen, Dorothy and Tom Amundsen, Rose and Art Raymond, Marge Holt, Teri and Bob Robb, Kathy and Mark Lowe, Joy and Dick Robb, Janice and Cecil Dunlop, Eva and George Naleznсy, Phyllis and Louis Hass, Sonya and Dick Ganyo, Lois and Kenneth Pantzer, Inez Matheson, Wally Nomeland, Bill Robb, Connie and David Lehrke, Cindy Lupien, Cher and Vernon Steele, Sue and Tim Brandreit, Becky and Rick Odden, Lois and Steve Atchison, Nona Hellen, Leslie and Juston Anderson, The Rev. Floyd Miller, Pam and Bruce Jacklitch.

Other generous donations were made from these friends and family in memory of their loved ones:

Sylvia Gates from John Gates, Kathy and Sylvester Beishir, Pat and Butch Whelan, Kay Bernardo; ***Bettye Dyer*** from Joe Dyer; ***Arlene Yager*** from Donna Pagano; ***Millie O'Toole*** from Donna Pagano; ***Paul Sheehan*** from Gloria R Sheehan; Patricia Jeans from Rose Jeans; ***Helen Smuczynski*** from Joan Spehar, Elaine Smuczynski; ***Cornelia M. Domreis*** from Oliver J Domreis; ***Vincent Kuklewski*** from Carol Kuklewski; ***James W. Donovan, Jr.*** from Nora A Motherway, Maureen, Elaine, Annie, Kathy and Michelle; ***Anthony Dean Puccia*** from Mary Puccia and co-workers at Regeneron Pharmaceuticals, Inc.; ***Carolyn Platteter*** from Gary Platteter; ***David Jones*** from Lois M Williams, Theresa Cipiti.

Please accept our sincerest sympathy over the loss of your dear loved ones. Thank you for your kindness in choosing the APF to honor their memory. Your generosity helps the APF enhance the health of people with porphyria and helps enhance the education of those doctors who have the responsibility of treating them. We appreciate your kindness at this difficult time.

DNA TESTING UPDATE

Many of you have asked for an update on the DNA testing. The APF-DNA Testing Laboratory at the Department of Human Genetics at The Mount Sinai School of Medicine of New York University under the direction of Drs. Robert J. Desnick and Kenneth H. Astrin currently offers DNA-based testing for Acute Intermittent Porphyria (AIP), Congenital Erythropoietic Porphyria (CEP), and familial Porphyria Cutanea Tarda (fPCT). Gene testing for Hereditary Coproporphyria (HCP), Variagate Porphyria (VP), and Erythropoietic Protophyria (EPP) will become available in 2007. The cost of identifying a family mutation is $500 and then $100 to test each family member. Typically, DNA testing takes 3 to 6 weeks and requires only a small amount of blood shipped overnight at room temperature. Once the lesion is detected in an affected person, other members of their family can be tested by simply collecting cells from inside your mouth using a kit provided by the laboratory or sending a small blood sample. If you would like to have DNA testing, APF members should contact Dr. Kenneth Astrin at 212-659-6783 or Kenneth.astrin@mssm.edu.

Previously, DNA testing by the Mount Sinai Porphyria DNA Testing Laboratory required bichemical evidence of porphyria. Now, testing is offered without this requirement to all patients. Of course, it is preferred that a patient provides biochemical evidence (enzyme, and/or urinary ALA, PBG and porphyrin tests), although porphyrins and porphyrin precursors may only be abnormal when the patient is symptomatic. Biochemical testing can be performed by Dr. Karl Anderson's laboratory at the University of Texas Medical Branch in Galveston, kanderso@utmb.edu or 409-772-4662 or by several commercial labs.

Many of our members have asked why there is a need for the DNA tests if their porphyria has already been identified through biochemical tests. One reason is to confirm a biochemical diagnosis and the second reason is for family studies. Once your DNA test has been performed and your mutation identified, the identification of other affected family members, even if they are asymptomatic, can be easily and accurately determined. These family members are at-risk for developing porphyria symptoms, and once identified can be counseled to avoid certain drugs, dieting, and other precipitating factors. It is now clear that most families have different gene defects (or mutations) causing the defective enzyme. Thus, in each family, their specific gene defect can only be identified by DNA sequencing. Once the specific gene defect is known in a porphyria patient, other family members can be accurately diagnosed from a small blood sample.

In general, the gene tests will identify 97-99% of the genetics lesions that cause a particular porphyria, so these tests are quite reliable, and particularly useful in patients who are asymptomatic or who do not have clear biochemical confirmation of their porphyria. In each porphyria, there are specific gene defects that result in decreased enzyme activity and the accumulation of the porphyrin precursors, ALA and PBG, and /or specific porphyrins. For example, in AIP, the level of the enzyme, porphobilinogen deaminase, is half normal, and ALA and PBG accumulate, particularly during an acute attack. The reduced enzyme activity is due to the presence of a mutation in one of the two copies of the gene that codes for porphobilinogen deaminase. To date, over 250 different mutations have been found in AIP families. Similarly, in familial PCT, the level of the enzyme, uroporphyrinogen decarboxylase, is half-normal and a series of specific porphyrins accumulate in the urine, particularly when patients are symptomatic. Over 70 different mutations have been identified in the uroporphyrinogen decarboxylase gene in patients with familial PCT.

DNA testing is more complicated than biochemical testing and requires more time than measuring the levels of the enzymes or porphyrins. As mentioned above, most mutations are found in only one or a few families, so the entire gene involved in the suspected porphyria must be sequenced in each patient. However, once the specific gene defect is identified in a patient, all family members can be easily tested to determine who has the porphyria-causing gene defect.

A Member's Perspective: **Judy Kitchens** an APF member from California, describes her experience with DNA testing.

A deep and permanent gratitude is how the great majority of us should feel for the founders of the APF and for the person who has anonymously donated the funds for the development of the APF at Mount Sinai School of Medicine (MSSM) in New York. An individual's preliminary biochemical diagnosis DNA Laboratory is reliably confirmed by a DNA test for one of the defective enzymes.

Last year, I had the privilege of contributing a sample of my blood to assist in establishing a DNA test for porphyria variegate at the Human Genetics Department at MSSM. This entailed an initial screening which involved filling in a short form and sending it, along with previous test results, to Dr. Kenneth Astrin at the APF DNA Laboratory in New York. When they accepted me as a research subject, I had a small amount of blood drawn and mailed along with my brief family tree by overnight carrier to the laboratory. In exchange, at long last, I know without a doubt that I have the mutation and have shared the information with my family. I will always be very grateful that I was in a worthwhile research study that will benefit others for generations to come. Studies of this kind are essential for improving and even saving the lives of patients with acute porphyria. While I was reading about past clinical trials, I wondered why some of their helpful conclusions had not been put into practice. Then I realized sadly that change often requires money as well as time, and probably the financial wherewithal was not available. The research studies we financially support and participate in will determine the kind of treatments we receive. They will also allow us to become a part of medical history.

Read the rest of Judy's Account at the Patient Stories Section of the APF website: www.porphyriafoundaiton.com

Dr. Steven Shedlofsky

A St. Louis native, has been at the University of Kentucky (UK), College of Medicine Division of Digestive Disease and Nutrition in Lexington, Kentucky for 23 years, after spending three years studying porphyria with experts, Drs. Bonkovsky and Sinclair. He met his lovely wife, Karen there and together they have daughter, Lydia. Dr. Shedlofsky is the Chief of Gastroenterology at the Veteran Affairs Medical Center and Associate Director of the General Clinical Research Center, where he has developed a training program and curriculum for training junior faculty and graduate students in modern clinical research. He is a professor at UK and is board certified in Internal Medicine and Gastroenterology with clinical expertise and interests in Hepatitis B, Fatty Liver, Hepatitis C, Liver Transplantation, Disorders of iron metabolism and porphyrias and porphyrin metabolism. Dr. Shedlofsky's research interests are: basic mechanisms of injury in Hepatitis C, Hepatic cytochrome P450 enzymes, Drug metabolism, Iron and oxidative stress and porphyria cutanea tarda.

When his father died of coronary disease and a geneticially linked illness arose in his own family, Dr. Shedlofsky said, *"Having a genetic basis for my medical disorder makes me appreciate more the genetic disorders the porphyrias represent. The advances made in understanding coronary disease should give patients with porphyria hope that scientific understanding will improve their quality of life, too."*

As a board member of the APF, we have referred many patients to him. He recently told us, *"Evaluating and helping manage patients with porphyria is still a special treat for me. With so few physicians who understand these rare metabolic disorders, I not only feel a special pride in knowing about them, but also an obligation to use my training. Being in Kentucky in a relatively small city like Lexington, there aren't very many patients from the area who have porphyria. So when I do see patients, they've usually had to travel quite a distance. I try to make sure that my clinic team gets all the information I'll need, including the proper collecting and handling of urine and stool specimens. I then advise patients to have a close relationship with a caring local physician with whom I can correspond. I don't know if this is the best way to help. But it's worked for me and I hope it's worked well for the patients with porphyria whom I've had the privilege of meeting."*

His patients are very complimentary about Dr. Shedlofsky's kindness, brilliance, bedside manner and expertise in porphyria. We want to extend our thanks to him for his decades of service.

Lyon's Share

You may not know that our work at the APF is a labor of love; I have porphyria. Lelia has porphyria. Yvette has a family member with porphyria. Carol has a close friend with porphyria, and Elizabeth has a son with a rare disease. Thus, identifying with your hardships serves to reinforce our commitment to do our best to help those of you who are in need. However, we are not the APF. We are merely the ones who try to implement the services and programs that improve our lives. So when you see the acronym APF, remember the APF is you and me.

I would like to take this opportunity to explain to you what I think are our most important needs. Of course, we would love nothing better than to find a cure and be part of funding research for a cure, but we do not have the kind of funding it takes to accomplish this. However, there are on-going research projects and potential research projects that are the building blocks to discover a cure, but these projects need more funding. I know you are tired of everything needing money. I am, too. But believe me, you may be well now but then again, like me, it could happen that your well world collapses and you find yourself in a swell of physical problems. ***Research is the Key to Our Cure.***

Also, as discussed previously, we are in very great need of training doctors to be the future porphyria expert, which is why we established the ***Protect the Future*** fund. One family has given a great gift for you and me. They have funded the training for a young doctor, who is in training now to become a specialist. But three doctors in the entire country will be enough. Your donations are needed to help train more young experts.

We need to increase our ***Physician Education Program***. With targeted plans, we made a major impact in the areas of improved diagnosis and treatment, but there are thousands of other doctors who need to learn more. We want to enlarge the program to include primary care physicians. Together we can accomplish all of these important services and programs. **I can't do it alone. Let's do it together.**

Thank You

for participating in
National Porphyria Awareness Week.

It is not too late to help in your area. If you would like to help, we have suggestions on what you can do and supplies for your efforts.

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

The IN TOUCH Network and Meetings

By joining the IN TOUCH network, you will receive the names and contact information of other IN TOUCH members in your area. You can also check the time and location of next IN TOUCH meetings on the APF website. To host an INTOUCH meeting, contact the APF.

Physician Education

The APF has expanded the Physician Education section of the website. If you have not told your doctors about this special section just for them, please encourage them to take a look. They can also take the Continuing Medical Education Course for fee CME credit on that same section. Also, if you want your doctor to receive a comprehensive packet of information, please contact the APF at Elizabethapf@aol.com or 1 866 APF 3635.

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Address service requested

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