



## **MERRY CHRISTMAS, HAPPY HANNUKAH, HAPPY HOLIDAYS, HAPPY NEW YEAR**

*Yvette, Jessica, Carol, Amy, Susan, Desiree, Lucy and Webmaster Rob have enjoyed serving you and your families throughout this past year. We pray that the love of God abound in your hearts and minds during this wondrous season and may your lives flourish with love and joy. Please know that each of you are important to us at the APF and that we are here to help. Contact us when you have a porphyria need, question or need a caring friend.*

### **CINDY ROY, PhD** provides scientific oversight of the Porphyrias Consortium.



Cindy Roy, PhD, a program director at the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK) at the National Institutes of Health, first realized she was interested in molecular biology in 10th grade.

"I had always been curious about the sciences," explained Roy, now a molecular hematologist. She wrote an extended essay about genetic engineering during her senior year of high school, and then earned a bachelor's degree in biochemistry from the University of Maryland, Baltimore County, followed by a PhD in cell biology from Oregon Health & Science University.

Prior to joining the National Institutes of Health (NIH), Roy's independent research program studied the biological function of proteins that influence red blood cell production. In her current role, Roy is responsible for overseeing the Porphyrias Consortium (PC), led by Robert Desnick, MD, who is a member of the American Porphyria Foundation's (APF) Scientific Advisory Board and is also a professor and chair emeritus, and Dean for Genetics and Genomic Medicine at the Mount Sinai School of Medicine in New York City. The consortium aims to use high-quality observational and interventional studies to improve the health of people who have porphyria. The PC is a part of the Rare Diseases Clinical Research Network, a network of consortia that is an initiative of the National Center for Advancing Translational Sciences, also at NIH.

The PC is a joint effort by the APF, the PC investigators, and the NIH. In her position, Roy oversees the PC closely to establish priorities for projects, to refine observational studies and clinical protocols, and to solicit the expertise of NIH colleagues to support the goals of the consortium.

When she's not working, Roy enjoys gardening, as well as hiking and kayaking with her family. Her career path has been a fulfilling one, Roy said. She appreciates and enjoys cultivating partnerships with the APF, the PC investigators, NIDDK colleagues Averell Sherker, MD, FRCPC, and Edward Doo, MD, and colleagues in the Office of Rare Diseases Research at the National Center for Advancing Translational Sciences, also part of the NIH.

She is grateful for the APF's leadership within the PC, and she expects the PC will continue to lead progress toward novel treatments and improved outcomes for people who have porphyria. *Editor's note: Dr. Roy has been very helpful to both the PC and the APF. We sincerely appreciate her insight, suggestions and interest.*

### **Comprehensive Packets for You and Your Doctor** are available upon request.



It is important to find a physician who is knowledgeable about porphyria. If you would like your doctor to learn more about porphyria, the APF's doctor packet is the best way to accomplish educating your physician. All of the educational material within the packet is written by porphyria experts. Below are publications that are included in the APF's comprehensive doctor packet for acute porphyrias. It is easy to request. Just contact the APF, provide your doctors' names and addresses, and we will forward this outstanding packet to your doctor the same or next day. In addition, we also have a patient packet for you. The list below notes the components included in each packet. If you have more than one doctor, we are happy to send a packet to each one. If family members need a packet, call 713.266.9617 or Toll Free 866.APF.3635.

#### **Physician Packet**

*Annals of Internal Medicine*

*"Recommendations for the Diagnosis and Treatment of the Acute Porphyrias"*

*Management of Acute Porphyria* brochure

*Panhematin* brochure

CME course brochure

Safe/unsafe drug list URL

Emergency Room Guidelines

#### **Patient Packet**

*Overview of All Porphyrias* brochure

*Panhematin* brochure

APF Newsletter

APF membership form

Patient letter from Desiree Lyon

How to become an APF Member

Wallet Warning Card

*Porphyria Live* DVD

## **FDA EPP MEETING** Hankies were in order during the six hour gathering at the FDA. Tears flowed freely as



The sunless Warriors Club

Olivia D

The FDA meeting

Cassie and Mitchell H

Sue M

old and young alike shared their EPP experiences with each other and the FDA Dermatology Committee. The weekend began when EPP patients and their families gathered at the Holiday Inn in College Park, MD on Oct 23, 2016 for a pre-meeting for the **Patient Focused Drug Development** meeting on October 24 at the FDA. Most of the attendees were EPP patients, but numerous family members and friends came for support. The pre-meeting provided EPPers an opportunity to engage others with EPP, and for some, it was the first time they had ever met another EPP person. It was especially fun for people who had met via Facebook to meet finally face to face.

The next morning the attendees arrived at the enormous FDA White Oak campus. The EPP meeting there was the largest gathering of its kind ever held at the FDA. Over 100 EPP people and their families and friends gathered in the dimly lit conference room to explain EPP and share their experience with an FDA panel, including Dr. Kendall Marcus, who heads the Dermatology Committee. This particular committee will be the regulators who will hopefully approve the revolutionary treatment, Afamelanotide/Scenesse.

The meeting began with presentations by Dr. Henry Lim, who explained EPP as a disease. He was followed by Dr. Joyce Teng speaking on treatments and then for the special part of the event, there was a panel composed of EPPers, Monica Fleegel, Madelyn Havard (11), Victor Mejias, Kerry Wiles and Meghan Rohn (16). In four minute speeches, they conveyed moving testimonies about EPP that left few dry eyes among the attendees. When it was time for the EPP attendees to add to the comments, the stories were powerful, poignant and formidable. They spoke of the inferno pain of EPP, the mechanisms they used to avoid the searing light and the subsequent isolation and the depression that occurs in the life of an EPP adult and child. Although the stories were heartbreaking, more importantly they exemplified the courage of every person who suffers from EPP.

The afternoon consisted of FDA presentations and a panel of experts, Dr. Manisha Balwani, Dr. Robert Desnick, Dr. Henry Lim and Dr. Maureen Poh. Their comments were profound, not only in the science they presented, but in their clear understanding of the biochemical mechanisms of the disease and compassion for what an EPP person endures in their lifelong struggle to adapt. Lastly, the EPP community closed the meeting by answering questions from the FDA and sharing concerns about their future, their need for a treatment, and their almost normal life when they had the Scenesse implant. Shawn Willis said that while he was taking Scenesse, he could even do his missionary work in Africa without any fear of a serious reaction. Mike Kenworthy spoke about the many clinical trials he had joined over the past 69 years and added that NONE had worked until Scenesse. Rob Saupe commented that his summer on Scenesse was the best summer of his life, but it was followed by the worst summer of his life because he no longer had Scenesse. Fortunately, there was an ongoing webcast so people around the country could have a bird's eye view of the proceedings.

Jessica Hungate, Director of Patient Services at the APF, who arranged the patient pre-meeting, hotels, transportation, etc., deserves our heartfelt thanks. Thanks, too, Tommy Nguyen, producer of *NBC Dateline*, for filming the event. We hope his film will be used if the FDA finally approves Afamelanotide. His *Dateline* program brought EPP to the attention of the country. Post your comments to the FDA here until December 24, 2016: <https://www.regulations.gov/document?D=FDA-2016-N-1493-0001>.

## **DON'T FORGET YOUR LIVER SCANS** Experts suggest that people with acute porphyrias and PCT



have an annual Liver Scan. Below is a list of articles relating the high incidence of Hepatocellular Carcinoma and porphyria, as well as a list of individual articles.

[https://scholar.google.com/scholar?q=hepatocellular+carcinoma+porphyria&hl=en&as\\_sdt=0&as\\_vis=1&oi=scholar&sa=X&ved=0ahUKEwi3h\\_aLjYHQAhWGbYKHf1iCgAQqQMIODAA](https://scholar.google.com/scholar?q=hepatocellular+carcinoma+porphyria&hl=en&as_sdt=0&as_vis=1&oi=scholar&sa=X&ved=0ahUKEwi3h_aLjYHQAhWGbYKHf1iCgAQqQMIODAA)

### **Hepatocellular Carcinoma in Patients with Acute Intermittent Porphyria**

<http://onlinelibrary.wiley.com/doi/10.1111/j.0954-6820.1984.tb05005.x/full>

### **Acute hepatic porphyria and hepatocellular carcinoma.**

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2246695/>

### **Porphyria Cutanea Tarda and Hepatocellular Carcinoma:** Frequency of Occurrence and Related Factors

<http://www.sciencedirect.com/science/article/pii/S016882788580746>

### **Hepatocellular carcinoma in porphyria cutanea tarda:** Frequency and factors related to its occurrence

<http://onlinelibrary.wiley.com/doi/10.1111/j.1600-0676.1992.tb00557.x/abstract>

## PCT AND HEP SUMMARY AND RECOMMENDATIONS



- Porphyria Cutanea Tarda (PCT) and Hepatoerythropoietic Porphyria (HEP) are cutaneous porphyrias caused by reduced activity of the heme biosynthetic enzyme uroporphyrinogen decarboxylase (UROD). PCT can be inherited or acquired (approximately 20 and 80 percent of cases, respectively). In both familial and sporadic disease, acquired susceptibility factors such as alcohol use, smoking, and hepatitis C virus (HCV) infection are common. HEP, which is exceedingly rare, is due to mutation of both *UROD* alleles. PCT generally presents in adulthood; HEP can present in children or adults depending on the degree of UROD deficiency.

- Typical features of PCT include chronic blistering photosensitivity, especially on the backs of the hands and other sun-exposed skin, which can lead to infection, scarring, hyper- and hypopigmentation. Many patients have elevations in serum transaminases, but neurovisceral attacks do not occur. Plasma and urine total porphyrins are elevated, with a characteristic pattern. Manifestations of HEP are similar, but more severe, and often develop in early childhood.

- As with other porphyrias, the evaluation for PCT is done in a stepwise fashion, with measurement of plasma or urinary total porphyrins as first-line tests. If positive, this is followed by a more extensive testing algorithm.

- The diagnosis of PCT is made by documenting the associated biochemical pattern of porphyrin increases, which includes increased plasma or urine porphyrins with a predominance of highly carboxylated porphyrins. Plasma peak fluorescence at 620 nm distinguishes PCT from variegate porphyria (VP); total fecal porphyrins in PCT may be normal or elevated with a characteristic pattern that includes isocoproporphyrins. Molecular analysis shows a *UROD* mutation in approximately 20 percent of cases; this is not required for diagnosis. HEP is diagnosed by these same porphyrin increases, which are often more marked than in PCT, and a marked elevation of erythrocyte zinc protoporphyrin. HEP should be confirmed by molecular studies that demonstrate biallelic *UROD* mutations.

- The differential diagnosis of PCT includes other blistering cutaneous porphyrias, other phototoxic skin disorders including pseudoporphyria, and other causes of urinary or plasma porphyrin elevations.

- All patients with PCT who have active skin lesions should be treated with phlebotomy or low-dose hydrochloroquine, as well as reduction of susceptibility factors. The choice between phlebotomy and hydrochloroquine depends on the degree of iron overload and other susceptibility factors. A full discussion of the management and prognosis of PCT and HEP is presented in detail separately.

Read about PCT on the APF website: <http://www.porphyrifoundation.com/about-porphyrina/types-of-porphyrina/PCT>

Read about HEP on the APF website: <http://www.porphyrifoundation.com/about-porphyrina/types-of-porphyrina/HEP>

**MICHELE LUDEWEG** *I was diagnosed with Porphyria Cutanea Tarda (PCT) in 1983 at age thirty seven. I had always had horrendous skin and infections of the skin. Within twenty-four hours of sunbathing, I broke out in patches of blisters on my forearms. This was the first symptom. But for a long time before this, my hair had been growing much faster than normal and my fingernails would split and crack off the top layer. My face had developed a brown raccoon mask. But I didn't feel ill. These blisters intensified with more sun exposure and broke open into sores which wouldn't heal. My skin was so thin that the slightest bump would tear a piece of it off and then another sore to heal. It took over eight months to get it under control the first time. I had to get really pushy with my family doctor to visit a specialist, but when I did, he was a gem and was familiar with porphyria. God bless him. We have a list of do's and don'ts that have worked pretty well.*



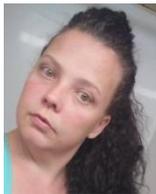
*I came out of remission two more times, the last being in 1990. I have had some other odd and unexplained illnesses and experiences and hope to get to the heart of it all as time goes by. Blessings to all of you.*

**MEMBER STORIES** The APF website [www.porphyrifoundation.org](http://www.porphyrifoundation.org) has a lengthy list of all types of patient stories, like Michele's. Read about YOUR TYPE: AIP, VP, HCP, PCT, EPP, XLP, AND CEP.

<http://www.porphyrifoundation.com/about-the-apf/member-stories>

We need your story, too. The most frequent comment we hear from newly diagnosed patients is how much it meant to read the experience of other people's hope with the same disease. Your story, and those of other people with porphyria, are very important because sharing your experience gives people hope that they will improve and lets them know they are not alone. So please consider writing your experience or contacting Desiree, Jessica or Yvette at the APF office, and share it with them. They will write it for you and post it on the APF website in the *Member Stories* section under your type of porphyria. Please contact the APF: [porphyrus@porphyrifoundation.com](mailto:porphyrus@porphyrifoundation.com).

**MEDICAL HEROS** **Amanda Rich** is the proud mom of Suzy and Sophie, and wife of Adam. Amanda serves as a greeter for the APF Facebook groups. Recently, she volunteered for the research program at the Galveston, Texas Porphyria Center, which is a long way from her Boston home. When she arrived, she met two other AIP patients in clinical trials as well as Dr. Linda Ede and study director, Dr. Karl Anderson. Having never flown before, for Amanda, it was a new adventure and a personal sacrifice. Amanda has had quite the porphyria journey. To stop her persistent attacks, she receives Panhematin, but has had to have her port replaced six times. *Thanks Amanda!!!*



**Nichol Kirby** is another "Medical Hero." Aside from hosting a recent Patient Education Meeting in her hometown of Indianapolis, IN, Nichol also flew to Galveston for the Panhematin study shortly after. Nichol is a cheerful young woman who lends a lot of support and camaraderie on the APF social media groups. She is working on her Master's degree, which is quite an achievement for a person with frequent AIP attacks. Fortunately, her sister Monica is always by her side helping Nichol through the rough patches.



**Mary Schloetter** travelled to Galveston from San Diego to participate in the Panhematin trials. Also, Mary's in-depth study of acute porphyria has enabled her to provide some of the most interesting and educational comments on social media. Mary has AIP which allows her to more fully understand the suffering of other acute porphyria patients. This is important for issues like chronic pain, misdiagnosis, etc. Thank you, Mary, for being a "Medical Hero."



You are needed to volunteer for research, including the simple Longitudinal Study that only requires you donate your blood and your time to answer a lengthy questionnaire.



**SUPPORT RESEARCH. IT IS THE KEY TO YOUR CURE!**

**PRODROME** A prodrome in medicine is an early symptom or set of symptoms that may indicate the start of a disease before specific symptoms occur. It is derived from the Greek word *prodromos*, meaning "precursor." Prodromes may be non-specific symptoms, or in a few instances, may clearly indicate a particular disease, such as the prodromal aura that often occurs before a migraine headache. In acute porphyrias, people have reported prodromes before attacks like, anxiety, chest pain etc. Amber Leighlynn has put her prodrome in her poem:

A pin prick...A tingle;  
An overwhelming feeling;  
That reality....is unreal.  
A small but subtle twinge,  
A spasm in my eye,  
My thoughts begin to race;  
My soul begins to cry.  
Porphyria stands by smiling;  
An evil, gleeful grin.  
As I begin to hold my breath;  
"Oh here you are again."

Do you have a prodrome, a warning sign that occurs prior to an attack that allows you to know an attack is imminent?

If so, what happens? Are your prodromes the same each time?

What actions do you take to prevent an attack when your prodrome occurs?

Have you always had a prodrome before an attack or is this a new occurrence? Contact 866.APF.3635

**RESEARCHERS ARE NOT ALL ABOUT SCIENCE (keep the carbs coming)** People with the



acute porphyrias, AIP, HCP and VP, need a large amount of carbohydrates during attacks. Eating healthy carbs, like honey, is very important. Congratulations to porphyria expert/researcher, John Phillips, PhD, at the University of Utah, for winning the first place award for producing Utah's best honey. As a beekeeper and now an award winning honey farmer, this porphyria expert is keeping porphyria people, and now bees, alive and well. You may have all read of the drastic decline in the number of bees. Without bees and other pollinators, we wouldn't be able to eat much of the nutritious and delicious food we eat on a daily basis. Thus, bees worldwide need our protection. Take action in your own backyards by planting a pollinator-friendly habitat, ideally free of pesticides so that it's the most nutritious and safe habitat pollinators can get or pass pollinator-friendly resolutions or policies at the city, university and school district level that are essentially working to do just that — plant safe, pollinator-friendly habitats. Have your good carbs handy.

**Thanks Dr. Phillips!! Keep your great research and great carbs coming!!!!**

**PANHEMATIN'S NEW WEBSITE** is a must see. Take a look at [www.IsItAIP.com](http://www.IsItAIP.com). Recordati Rare Diseases Inc. launched a terrific new website as a part of their disease awareness program for Acute Intermittent Porphyria - "Is It AIP?" The goal of the website is to help people who have symptoms that could be attributable to AIP to "Put a Name to the Pain." The website includes information about symptoms, diagnosis, and how to connect with the APF. There is also a symptom checklist that can be downloaded and used in discussions with your doctors.



The website also has a section for healthcare professionals who are looking to learn more about recognizing symptoms, diagnosing, and testing for AIP. This new website replaces the previous RRD disease awareness website for Healthcare Professionals: [www.diagnosingAIP.com](http://www.diagnosingAIP.com).

You will love the graphics, check out the symptom check list, and show your doctor, because YES, it may be AIP!! Many physicians have missed porphyria thinking that porphyria is rare and that your ailment couldn't possibly be porphyria. That is why it is important to show doctors websites like [www.IsItAIP.com](http://www.IsItAIP.com) and to order a free doctor packet from the APF. **Don't forget that Dr. Karl Anderson is conducting very important Panhematin research. Volunteers, who have frequent attacks, are greatly needed for this study. All travel expenses to and from the Galveston, Texas Porphyria Center are paid.**

**NEXT ALNYLAM TRIALS BEGIN IN 4 SITES** Patients with Acute Intermittent Porphyria (AIP) are travelling from all parts of the country to four of the Porphyria Centers, University of Alabama, University of Texas Medical Branch, University of California San Francisco, and Mount Sinai to participate in the newest Alnylam Pharmaceutical trials with their new RNAi treatment.



The premise is that hormonal changes, exposure to certain drugs, or dieting in AIP patients can cause the induction of aminolevulinic acid synthase 1 (ALAS1), the rate-limiting enzyme in the heme biosynthesis pathway, leading to the overproduction of aminolevulinic acid (ALA) and porphobilinogen (PBG) that trigger the attack symptoms. They reported positive initial results from the ongoing Phase 1 clinical trial with ALN-AS1, an investigational RNAi therapeutic which targets aminolevulinic acid synthase 1 (ALAS1) for the treatment of acute hepatic porphyrias. Importantly, ALN-AS1 was found to be generally well tolerated with no clinically significant drug-related adverse events to date. This data provides human proof-of-concept for ALN-AS1 as a potential therapy for AIP and other acute hepatic porphyrias.

Recently, Alnylam held a company fundraising event at their Boston headquarters that benefitted the APF. They invited acute porphyria patients, Colin McEwen, Amy Chapman and Rose Jeans to share their porphyria experience and the value of treatment for their attacks.

**PATIENT EDUCATION MEETINGS** Longtime APF member, **Ruth Dee Bruno** (3<sup>rd</sup> from left), hosted a patient gathering in the Chicago area. Twenty plus patients and APF members attended and shared their own stories.



**Nichol Kirby** hosted a meeting in Indianapolis. Nichol said she had a very congenial group who enjoyed meeting other porphyria people. Desiree joined the meeting by telephone to answer the many questions. Although not an MD, Desiree was able to field questions by answering from materials that the experts had published.



**Evelyn Jacobucci** hosted a meeting in Denver, Colorado on November 18<sup>th</sup>. Evelyn, her husband Sam, and their four kids have been active with the APF for a long time and often reach out to support other patients. Attendees enjoyed meeting one another and sharing their life with porphyria. Watch the APF ENEWS for details of the next meeting near you. If you are interested in hosting a meeting in your area, contact Jessica or Yvette at the APF office. The APF provides educational materials, DVDs and other information for patient education and support.

If you are not receiving the APF ENEWS, contact the office and give them your email address so that you can be added to the weekly ENEWS. You might also become a member of the APF at the same time. Joining is free, but we certainly need and appreciate every donation to the APF.

We would like to host meetings in Atlanta, GA; Santa Rosa Beach, FL; New York City, NY; San Diego, CA; Seattle, WA; Miami, FL; Birmingham, AL; Wake Forest, NC; Salt Lake City, UT; New Orleans, LA; and any city with a porphyria family. The APF will provide the supplies, but we need you to be the HOST. Please contact the APF if you are willing to help us arrange a meeting in your community. You will enjoy the camaraderie and learn a great deal about porphyria in the process. Thank you.

## **PLEASE SUPPORT THE APF**



For over thirty years, the APF has been the mainstay and only source for porphyria education and awareness for patients and physicians, research, government funding, NIH and FDA proceedings, as well as an array of patient support services that has a Scientific Advisory Board of renowned porphyria experts. The APF educational materials are all award winning publications, including the website, newsletter, E-NEWS, brochures, and ER kits. Patients and Physicians worldwide seek the APF's information, because they can be assured that the information is reliable. Misinformation about porphyria is prevalent on the internet, so it is essential to have reliable information that is written by internationally renowned porphyria researchers and clinicians.

You have made the APF such a successful and helpful resource for patients, their families and physicians. Thank you for your support to make these services and programs possible. We need your help to reach more doctors to provide them with updated data on diagnosis and treatment. Together we can make a huge impact on the medical community.

The APF also has the *Protect the Future* program to train the next generation of experts. These young doctors are being trained by the present experts before they retire and their expertise is lost. The APF needs your help to keep this very valuable program on target. It is a costly endeavor to train young doctors, but they will be our only future source of help and expertise. **THE APF EXISTS TO SERVE YOU AND YOUR FAMILY, BUT THE APF NEEDS YOUR DONATIONS TO CONTINUE THESE PROGRAMS AND SERVICES. PLEASE PROVIDE YOUR TAX DEDUCTIBLE GIFTS IN THE ENCLOSED ENVELOPE.**

## **ANOTHER WAY TO SUPPORT THE APF – AMAZONSMILE**



Many of you purchase numerous items from Amazon, not just books. Now you can support the APF through the AmazonSmile program! Amazon will donate 0.5-0.8% of the price of your eligible purchases to the APF, at no cost to you. Please make the APF your choice of a charitable organization. Support porphyria research while shopping!

Note, this program to provide donations to the APF will ONLY be available to shoppers who visit Amazon via a special web address, namely, [www.smile.amazon.com](http://www.smile.amazon.com) instead of the normal [www.amazon.com](http://www.amazon.com) homepage.

It is easy and free! AmazonSmile is the same Amazon you know – same products, same prices, same service. Thank you for supporting us! Please follow the link: <http://smile.amazon.com/ch/36-4401266>.

## **MOO'VE IT IN THE MOONLIGHT**



Shawn Willis and family once again held a **Moo've it in the Moonlight** 5K and 1 Mile walk in Burlington, NC on November 5th. Shawn owns a popular Chick-fil-A restaurant. The race benefits the APF's physician and patient educational and awareness programs. The attendees received a Dri-fit Shirt, Overall & Age Group Awards, and Chick-fil-A Prizes. In addition to the race, Shawn and his wife, Lori, attended the EPP meeting at the FDA where he related his extraordinary experience with Afamelanotide. We sincerely appreciate all Shawn and his wife do for the APF and people with EPP.

## **JESSICA AT GLOBAL GENES**



I am Jessica Hungate, and I am the Director of Patient Services for the APF. In September, I had the honor of representing the APF at the Global Genes RARE Patient Advocacy Summit in Huntington Beach, California. This summit brings together rare disease patients, advocacy groups, and experts from all over the world. While I knew this conference would be an amazing experience, it surpassed all of my expectations. During the summit, I was able to attend a variety of lectures and seminars on different topics, but all related to rare diseases. The presenters ranged from genetic researchers to pharmaceutical companies to patient representatives and even included several parents of children with rare diseases. Since over 160 diseases were represented, I learned about many conditions that I had never heard about and in turn, I used the opportunity to teach numerous people about porphyria and the APF.

After the conference, I was invited to attend the RARE Tribute to Champions of Hope Gala. The Gala featured many important speakers, performances, and even a few celebrities! Many of the speeches highlighted the difficulties and successes of those learning about and living with their rare diseases, while achieving things they never thought were possible. We hear many inspiring stories every day at the APF, and it was uplifting to see so many people from different backgrounds coming together and caring about rare diseases. I am excited to deepen the connections I established through this Summit and look forward to collaborating and implementing new porphyria programs in the future. This conference has greatly enhanced my passion for the rare disease community, and I know the memories will stay with me for the rest of my life!

**WHEN IT IS NOT PORPHYRIA** The following is an important story, because it addresses when you discover you don't have porphyria. It is frustrating to think you have the answer to your illness and then find you are back to square one. We hope Rene's diagnostic adventure will help you in your quest to find the real diagnosis.



*Hi, My name is Rene' and I have a rare metabolic disorder. My life started with horrible colic. My mother says that she only worried if I wasn't screaming at the top of my lungs. My symptoms really should have been investigated starting at 6 years old. Fortunately, they eased a bit when my grandmother fed me carbs and NEVER made me wait to eat. Ah, the love of a grandparent.*

*When I was in the 4th grade, my mother took me to a specialist in Portland. This is when things really started to get bad. It's interesting how our bodies know what we don't need. I was started on meds and would refuse to take them but mother forced me to take them. I do not blame her as she had no idea they were bad for me. Then migraines started in the 5th grade; no noise, no light and no crying as it made the pain worse.*

*Next I began to have ADHD and at 13 years old had my first episode of stomach pain. I remember being curled up in a ball on the floor crying. My mother thought I had gas. This is also when I began serious reactions to skin products. Later my ex-husband thought that my "mood issues" were a hormone thing, so I began to look for answers and even admitted to my OB that I was not normal. He sent me to psych who told me that I had ADHD and prescribed another medicine that made me worse. My disorder continued almost to my destruction. In my early 30's, I was tested for lupus and cancer. When my son developed my symptoms, I was sent to my first geneticist who tested me for PKU, Myoglobin and Alkaptonuria. The second geneticist tested me for Porphyria and told me there was nothing other than Porphyria that caused urine to change color in the sun, which I discovered later was not true. My 3rd geneticist, Dr. Joanne Nguyen, was intrigued and tested my entire genome. Three months later she had an official name to my disorder. I am the first diagnosed symptomatic carrier in the world for Malonyl CoA Decarboxylase Deficiency with multiple CYP450 mutations.*

**Thus, if people truly want the answer to what is wrong with them, they have to accept what is not wrong with them** and search for a doctor that will help them find the true diagnosis. No, I do not have Porphyria. Life with a rare disorder is hard enough. Not knowing what is wrong makes it even worse. Giving up on finding the reality of the situation is NOT an option if you really want to live life to the fullest. Am I perfect now?? No, but my quality of life is much better. Most importantly, my son will never have to go through what I have been through, like doctors telling him he has a psych problem or trying to give him meds that make him sick. He has a much better chance at life than I did.

**PODCAST** Dr. Fiona Rahbar is a board-certified dermatologist, as well as a Fellow of the American Academy of



Dermatology. Dr. Rahbar has completed an undergraduate degree in Neuroscience and Behavioral Biology at Emory University. Subsequently, she studied at and graduated from Vanderbilt University School of Medicine and completed her dermatology residency at the University of Virginia, where she served as Chief Resident during her last year of training. Most recently, Dr. Rahbar is practicing tele-dermatology in New York City. Her newest project, aside from treating

patients, is to host a podcast on rare dermatology and skin conditions like the porphyrias. She will be interviewing porphyria people in her upcoming programs. Visit: <http://www.fionarahbarmd.com/our-podcast>.

**IN MEMORY AND IN HONOR** We sincerely appreciate friends of the APF honoring their loved ones with gifts in their names. We are grateful for the trust you have shown to fulfill the APF mission of heightening porphyria awareness and education. Thank you.



**In Memory** We send our sympathy to the family and friends who honored their loved ones with gifts to the APF. We join them in thanking you for their donations: Charlie Shock, Shanna Rodenbeck for **Nancy E Soeurt**; Wayne E Oas, Tressler Alumni Assoc. for **Robert Davenport**; Charlotte E Beck for **Elva McCoy Denger**; Bill and Mary Lou Rickert for **Gina Marie Opperman**; Stephanie Adler for **Melinda M Marcalo**; Diana Sabella, Rosemarie Gabriel, Augustine J Angelina, Jr, Linda Rosbert, Nicole Tunis, Gina Pretko, Lois Mauro, Paul Kalani, Kathy and Timothy Manna, Andrea and Joseph Strain, Lorraine DiPietrantonio, Patricia A Kellen, Marie and Vincent Passalaqua, Jean H Grebowich for **Carol A Rusnak**.

**In Honor** We also thank those who donated in honor of a friend or family member: Kathleen Angela Shiel for **Ralph Gray**; Vickie Lirman for **Debora Mary Kim**; Anne Johnson for **Candace Johnson**; Charlotte J Beck for **Sarah Nicole Sundblöm**; Eric Beale for **Daphne Beale**.