



AMERICAN PORPHYRIA FOUNDATION

3rd Quarter, 2019

APF LEADERSHIP TRANSITION



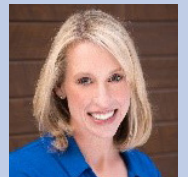
The American Porphyria Foundation now has over 11,000 members, which is a huge number from the two founding members we had at the initiation of the APF in 1982. Our staff consisted of me and Mr. Young, who has acted as President and Treasurer since day one. Our office was my kitchen table and my bedroom floor for mail sorting. My daughter, Lelia, and sister, Elizabeth, were our mail sorters and mail deliverers.

After three decades as Executive Director of the APF, I'm transitioning to Global Director. It was an exhaustive search to find the right person to take my position. Kristen Wheeden is that right person. Kristen, who has been working with the APF for two years, has surpassed our expectations. Since the APF was in my hands

since its beginning, I was overly cautious when choosing the new Executive Director. I'm confident that Kristen will bring the APF to new heights of service to patients and physicians. We all wish her well and thank her for undertaking such a challenging position.

~ Desiree

I am honored to have the opportunity to follow in Desiree's footsteps as your new Executive Director. We will work hand-in-hand as she transitions to Global Director, where she will realize her dream to use her expertise helping emerging organizations. As a porphyria advocate for the past decade and on the APF staff for the past two years - I am dedicated to each and every member of this extraordinary foundation. There is much good work to do!



~ Kristen, kristen@porphyriafoundation.org

APF GLOBAL PROGRAM

Since the inception of the APF, people around the world have contacted us for assistance. Our board of experts has consulted with doctors and the APF has provided information and support. When we began our new Global Program, the APF formalized our International Education Program. To date, we have assisted doctors and patients in 76 countries (listed below).

Our new additional agenda is to help patient groups form and grow support organizations. This includes helping them expand their member base and identifying group leadership, as well as doctors with porphyria expertise. We have trained physicians as experts and educated doctors around their countries in the porphyrias.

At present, we are working with new organizations in Mexico, the Middle East, Northern Africa, Chile and Pakistan. We have also become part of the Global Porphyria Advocacy Coalition (see right). Our goal is help each country worldwide to have a patient organization to help care for patients in their own respective countries!

Afghanistan, Angola, Argentina, Austria, Bahamas, Belgium, Belize, Botswana, Bulgaria, Chile, China, Columbia, Costa Rica, Cuba, Czech Republic, Ecuador, Egypt, El Salvador, England, Fiji, France, Spain, Belgium, Denmark, Australia, Chile, Brazil, Venezuela, Turkey, Oman, Saudi Arabia, Morocco, Greece, Guatemala, Honduras, Iceland, India, Indonesia, Iraq, Ireland, Israel, Italy, Jamaica, Japan, Jordan, Korea, Latvia, Luxembourg, Malta, Mexico, Monaco, Montenegro, Malaysia, Netherlands, Newfoundland, Nicaragua, Norway, Pakistan, Panama, Peru, Philippines, Poland, Portugal, Qatar, Romania, Russia, Scotland, Slovenia, South Africa, Sri Lanka, Syria, Tanzania, Thailand, Vietnam, Zambia



GLOBAL PORPHYRIA ADVOCACY COALITION (GPAC)

The GPAC has been established as an umbrella organization for porphyria advocacy and support groups worldwide, representing all types of porphyria. This organization will establish and maintain a global network of leaders, patients and physicians seeking to support all individuals impacted by porphyria and to safeguard their interest. The GPAC will provide a unified, collaborative voice for porphyria patients worldwide. The APF is a founding member of the GPAC and will continue to be intimately involved, representing our membership on a global scale.



DONATE TO THE APF TODAY!

porphyriafoundation.org or
1-866-APF-3635

A CHAT WITH DR. PHILIPPE WOLGEN, PRESIDENT, CLINUVEL PHARMACEUTICALS



Dr. Philippe Wolgen has dedicated fourteen years of his professional life on advancing treatment for Erythropoietic Protoporphyrin (EPP). As the president of Clinuvel Pharmaceuticals, he has a unique perspective on research and the challenges in bringing a novel treatment to patients.

On May 31, the US Food and Drug Administration (FDA) delayed their review of Clinuvel's drug Scenesse until October 6. Days later, when I met Dr. Wolgen, I expected he might be discouraged. Instead, he was fired-up and ready to meet this latest FDA challenge. Why? He explained that when this treatment finally meets with success in the U.S., it will be even more meaningful due to all the challenges that have been overcome. Persistence, combined with integrity, will have led the path to success.

After 16 years in medical school; Dr. Wolgen spent four years as a practicing physician, in academia, before pursuing a career that integrated his many talents. His approach to study and treatment is different. "It's good to be different," says Dr. Wolgen, "It's how you channel it that matters." Noting that people living with EPP must live their lives differently. He explains that, everyday challenges not only build character but make you different than others.

Before taking the helm at Clinuvel, Dr. Wolgen spent a year studying Clinuvel and their pipeline, originally named EpiTan. The technology and potential were there, and

an innovative leader may have an opportunity to bring it to fruition. An intriguing twist in the data captured his interest during his analysis of the company and presented a challenge. Typically, a treatment is tested for efficacy and safety. EpiTan had 20 years of safety data without efficacy data. This was the beginning of the development of Scenesse for the treatment of EPP.

Dr. Wolgen has spent years understanding this disease and the effect on patients. At the October 2016 FDA meeting in White Oak, Maryland, he listened quietly from the back of the room. Two moments affected his outlook on treatment for EPP. First, when a patient said, "I don't have the words" to describe an EPP reaction. Dr. Wolgen was struck that no common words exist for EPP patients and physician to characterize this disease. **"It is not pain. Pain can be managed by medication. It is beyond pain," he said.** Dr. Wolgen believes that until we characterize EPP appropriately, the impact of this disease on patients will not be understood.

The second moment, when a mother shared that her son "was changing before her eyes with the impact of this disease." The psychological nature of EPP intrigues Dr. Wolgen. He once witnessed a patient reaction, describing the pain as a "psychological decomposition." This happens when a patient is beyond pain. The patient had been outside in the sun with no shade, no cover and no way out. "It was like an animal in a cage, out of his mind writhing in pain," he said, "This is not 'stinging or burning', it's beyond words."

Dr. Wolgen firmly believes that the patient community – including the physicians who diagnose and treat them -- need to characterize the disease in more accurate way, "It is a chemical reaction, akin to radiation, boiling on the inside."

The approval of Scenesse in the U.S. and access to reimbursement in approved countries is in sight. "I wake up with a smile on my face every day," says Dr. Wolgen, "because the bonus, is that I know that this treatment works. We achieved the goal in science, and we have achieved the investment goals. It is true, it is genuine, it is safe, and it works."

He explains that if a drug can get "a wow," that's worthwhile. But if it can have the dramatic effect of *changing the quality of life*, then your goal is achieved.

If Dr. Wolgen is motivated by a challenge, the FDA approval is proving to be a great one. First, Scenesse was granted *Fast Track* designation. Then it was given *Priority Review*. For every step forward, there have been delays. Despite these challenges, Dr. Wolgen remains positive. "At some point, they will have no more reasons to deny and we will have a product in the U.S.," he said.

"You can do your best to succeed and you can do your utmost not to fail. Those are two separate concepts." We don't yet know the outcome in the U.S. for Scenesse, but we can be sure that Clinuvel will do their best to succeed, with Dr. Wolgen leading the charge. ~ Kristen Wheeden



Kids Corner

Kids have questions too! I asked three young APF *Shadow Jumpers* what they would ask Dr. Wolgen if they had the chance...

Question 1: How do you make it? (Scenesse)

Dr. Wolgen: Take an egg white (the protein), throw away the yolk, whisk it, coat it with sugar, roll it into a spaghetti shape, and cut the spaghetti into small pieces. It's a little bit like that.

Question 2: Can I have it? (referring to Scenesse)

Dr. Wolgen: I have a story. It's a little bit like Christmas shopping. My friend has three children. (Two girls and a boy.) Every year, their family goes shopping together for Christmas, so each could pick out one toy. Their little boy didn't want one toy – he wanted *all* the toys. He had a big tantrum. The next year they did it dif-

ferently. His parents said, he could get anything he wanted. The boy stacked up all his toys at the register. Then his dad said, "I let you buy all these toys, but I didn't tell you when you could have them." Like Scenesse. You can have it, but I can't tell you when.

Question 3: What do you do?

Dr. Wolgen: There is an old British comedian named Tommy Cooper. Tommy did tricks with magical sticks. He would put plates on top of all the sticks. Then he would spin the plates. He was so happy when all the plates were spinning at the same time. His smile was so big. Then one plate would start to fall, then another, and he'd have to get them spinning again. And he ran around doing that and it was very funny. That's what I do...I keep all the plates spinning.





SCENESSE UPDATE



CLINUVEL

Clinovel Pharmaceuticals Ltd. received notice on May 31 that the PD-UFA (Prescription Drug User

Fee Act) review date for Scenesse was delayed from July 8 back to October 6. The FDA will communicate either an approval or a Complete Response Letter (CRL) with an explanation of further information needed. This is yet another delay in the long process to bring Scenesse to EPP patients in the U.S. The APF is sending letters, information, updates, and patient stories to the FDA on a regular basis in support of approving this life-altering treatment. If you would like to send a letter, please reach out the APF for contact information.

#approvescenesse

PANHEMATIN® UPDATE



RECORDATI

Panhematin® will soon be available to patients with acute porphyria in Canada. Please stay tuned for further information. This life-saving treatment has been available in the United States for nearly forty years, and we are excited for our Canadian friends to have access to heme treatment.

GIVOSIRAN UPDATE



Anylam Pharmaceuticals, Inc. announced on August 5 that the FDA has accepted the New Drug Application (NDA) for Givosiran and granted it Priority Review. The FDA has set an action date of February 4, 2020 under the Prescription Drug User Fee Act (PDUFA). The European Medicines Agency (EMA) has already validated the drug's Marketing Authorization Application and it was granted accelerated assessment. An accelerated assessment can reduce the Agency's evaluation time by up to 60 days.

Givosiran is a RNAi therapeutic targeting aminolevulinic acid synthase 1 (ALAS1) in development for the treatment of acute hepatic porphyria (AHP). By reducing the accumulation of neurotoxic heme intermediates, aminolevulinic acid (ALA) and porphobilinogen (PBG), Givosiran has the potential to prevent or reduce the occurrence of severe and life-threatening attacks, control chronic symptoms, and decrease the burden of the disease. The safety and efficacy of givosiran were evaluated in the ENVISION Phase 3 trial with positive results; these results have not been evaluated by the FDA, the EMA or any other health authority.



WE HAVE NEARLY 12,000 MEMBERS IN THE APF DATABASE!

The database of the APF remains a critical way to communicate with our members, guide patients toward physicians familiar with porphyria, and alert members to research opportunities and regulatory updates. Privacy is maintained – no information is ever shared without consent. Contact the APF to add or update your information today. We'd love to hear from you!

ACUTE HEPATIC PORPHYRIA TESTING RECOMMENDATIONS

Members often ask how often PBG and ALA should be tested. Recommendations for Biochemical Testing in Acute Hepatic Porphyrias from our Scientific Advisory Board are as follows:

In patients with Acute Hepatic Porphyrias (AHP), signs and symptoms of acute attacks often include severe abdominal pain, nausea, vomiting, muscle weakness, seizures, and mental status changes. These manifestations are due to accumulations of the porphyrin precursors, aminolevulinic acid (ALA) and porphobilinogen (PBG). Biochemical testing of urine ALA and PBG is important in the diagnosis of AHP and should ideally be collected when a patient is symptomatic. A random sample is sufficient for testing, 24-hour samples are not necessary. Most patients with Acute Inter-

mittent Porphyria will have elevated levels of ALA and PBG even if they are not actively having symptoms. In Hereditary Coproporphyruria and Variegate Porphyria, these levels may decrease to normal in between attacks.

Levels of ALA and PBG can serve as biomarkers for disease activity and increase markedly during acute attacks. For accurate assessment, levels should be measured prior to the administration of Panhematin which is known to decrease ALA and PBG shortly after an infusion. As well, testing should be completed at a laboratory (such as Quest Diagnostics, UTMB and Sema4) that measures these levels expressed per gram or mmol of creatinine. This corrects for diluted urine, thus, minimizing false negative results. Small amounts of light exposure typically do not affect the levels of ALA and PBG. Measuring ALA and PBG levels around an acute attack, before and after Panhematin treatment, can also provide a

clinical picture of treatment response. However, it is not necessary to repeat this daily during every hospital admission or outpatient infusion. This is generally done based on the treating physician's judgement, and consultation with a Porphyria specialist is recommended. For patients with confirmed diagnoses, administration of Panhematin should not be delayed until ALA and PBG results are returned.

Patients should have ALA and PBG levels routinely monitored as part of their yearly follow up evaluation. This includes patients who are symptomatic (sporadic or recurrent acute attacks) and those who do not have symptoms but were found to have elevated levels of ALA and PBG on testing, referred to as asymptomatic high excretors (ASHE). In latent patients (those with a known genetic mutation but normal levels of ALA and PBG), repeat testing should be done as clinically indicated if the patient develops symptoms of AHP.

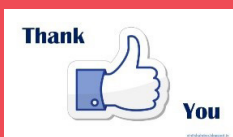


FDA RARE DISEASE LISTENING SESSION

The APF has secured a coveted RARE DISEASE LISTENING SESSION with the Food and Drug Administration (FDA), which will take place on August 26th. These sessions are intended to be a resource for the FDA to engage with patients or their advocates. It is an excellent opportunity to share our patient perspective with the FDA. Sessions help to inform regulatory decision-making

and educate review staff about rare diseases. Our FDA counterpart shared that our meeting request received the most interest of any other. Our objective for this one-hour session is to help the FDA understand the impact of living with porphyria, to differentiate the porphyrias and the burden of living with each disease and focus on our need for treatment for each type of porphyria!

ONE MILLION "LIKES" TO OUR APF FACEBOOK ADMINISTRATORS, MODERATORS, AND GREETERS!!



The APF Facebook groups are extremely active with new members, regular

comments, frequent questions and constant support. Thank you to the all our wonderful volunteers who moderate and manage under the leadership of Amy Chapman, Director of Social Media. You are appreciated!!

YOUR MEDICAL INFORMATION, YOUR LABORATORY RESULTS, AND YOUR PRIVACY!

At the American Porphyria Foundation, we aim to protect the health and well-being of all patients and members. To accomplish this, the APF will only provide you with information that has been written or approved by the porphyria experts on our Scientific Advisory Board. These physicians, along with your own medical professionals, are your resource for health information. We urge you to be vigilant about who you allow to see your medical information and to be careful about taking advice from fellow patients and other non-professionals. Make sure that you are not sharing your medical records and personal medical data with anyone other than your healthcare provider, especially on social media. Sharing your information with people other than healthcare providers can also compromise your identity. If you have questions about your lab results, please seek help from your healthcare provider. If any medical information is shared with the American Porphyria Foundation, it is immediately shared with members of our Porphyrias Consortium, as we are not medical professionals and do not interpret laboratory information. If you have any questions, please contact the APF via email at porphyrus@porphyriafoundation.org or via phone at 301.347.7166. Your health and privacy are important to us!



SHANA WILCOX AND AMBER 81 PROTECTIVE FILM

Heather & Tim Wilcox from Petrolia, Ontario knew there was something wrong when their daughter, Shana, had a severe reaction to the lighting from an incubator she had been put in shortly after her birth, but a diagnosis would not come for many years. Similar reactions to lighting continued throughout her childhood and after the family moved to a new subdivision with few trees to provide shade, her reactions continued to increase with her sun exposure. Shana's hands, feet and face would often swell, and she would complain that her skin felt like it was on fire and lava was being poured over her skin. Shana would also experience significant pain and swelling on her face after a simple dental cleaning. After years of referrals to specialists and no explanations to explain her symptoms, Heather came across an article on the internet on Erythropoietic Protoporphyrria that matched Shana's symptoms.

She brought Shana and a potential diagnosis – EPP – to the doctor. Though her physician thought it was an unlikely diagnosis due to how rare it was, the testing was done, and she was officially diagnosed at age 15 and confirmed via genetic testing with Dr. Bloomer, at UAB, whom helped them through this process. As the years passed, Shana could typically manage no more than 15 minutes of sun exposure and was also monitored closely with her liver functioning. This past May, a new challenge arose. At first thinking she was experiencing symptoms of the flu, a trip to the emergency room confirmed that Shana had experienced a ruptured appendix. This meant surgery! Due to the risk of internal burns from the operating room lamps, Heather went into gear, resourcing protective film for the surgical lights. Although the family had some protective film on hand over the years, there wasn't nearly enough to ensure all the lamps would be covered. Armed with educational information from the APF, Heather & Tim did NOT let them take their daughter into surgery until they were prepared to protect her. But, what did they need and where could they get it on short notice? Heather assembled a team ready and willing to support their daughter. The APF advised on film, Window Film Systems in London, Ontario provided specs for the Madico film, Amber 81 and confirmed they had it in stock and the surgical team and OR Department agreed to have it installed. Jake, and the Window Film Systems Team, took extra time to research EPP after being contacted and provided the family with very helpful information. He also insisted on delivering the film to the hospital so that Shana's family could remain with her. They also suggested the possibility of building a frame so that the film could better attach to the surgical lamps. The kindness of strangers willing to go the extra mile made all of the difference in a stressful situation. After the team of surgeons reviewed Shana's case, they decided to treat her ruptured appendix with IV and oral antibiotics to give them the extra time needed to do a risk analysis and ensure all precautions are put in place for surgery. Shana is doing well and on her road to recovery!

KURE4KAM



The APF is joining forces with Kure4Kam, a group created to support Kameron Kadinger, who lives Homozygous Acute Intermittent Porphyria. There have been fewer than ten known cases of this type of porphyria in the world. To be considered homozygous, each parent has to have the gene for AIP and then both have to pass it on to their child. Homozygous Acute Intermittent Porphyria presents differently than typical AIP and they are progressive. His symptoms are daily and Kam experiences additional debilitating effects such as developmental delays, seizures, apnea episodes and more from this type of porphyria. He received the first liver transplant in this disease, an experimental surgery with unknown benefits. Kure4Kam will remain a separate program under the umbrella of the American Porphyria Foundation, with access to considerable expertise and a home for program management. Funding raised by Kure4Kam will support critical research on homozygous AIP.



A note from Kam's mom, Amanda:

Kameron, or better known as Kam man, is a special 2-year-old. His 3rd birthday is around the corner, but he is still be little baby. He is a spunky and very opinionated little boy; I shouldn't forget tan and beautiful too. Yes, I said beautiful. He has piercing blue eyes and flawless complexion. His pictures can be misleading, he appears like any other typical 2-year-old. It is more common than not to hear people say that Kam doesn't appear sick but that couldn't be further from the truth.

Kam's journey with porphyria has taken us across state lines to find treatment for his progressive disorder. As his mother, I spend the majority of my interaction with strangers, family members, friends and health care workers just educating about homozygous acute intermittent porphyria. We have had ups and downs but overall, we have been met with empathy and kindness. Kam deserves love and it is my personal mission to make sure Kam feels loved every second of every day.

Kure4Kam is joining forces with the APF and I couldn't be more excited. This will allow for bigger and better opportunities to fund a Kure4Kam. We are all dying, it just so happens Kam is dying quicker than most of us. I am hoping through fundraising, networking and research we can improve Kam's quality of life. The APF has connections, compassion and drive to help all porphyria patients. I am excited to announce through our combining forces we will be attempting to collaborate with the top physicians across the world. So, hang tight and keep an eye out for exciting things to happen because I assure you, they will!

Porphyria is an umbrella of disorders that affects people in all sorts of ways. Kam's dad and I, Keith, have AIP. Lucky enough, we do not suffer from the attacks (I may have had one in my early adulthood, but I am unsure). It is important to remember that even the most well-known porphyrias are rare, but there is branch of ultra-rare porphyrias as well. Kam falls in the category of ultra-rare. He is developmentally at a 5-6-month level, the first homozygous acute intermittent porphyria to receive a liver transplant, nonverbal. He is completely dependent on myself and Keith to live and will continue to be for the remainder of his life. I will continue to fight for Kam's quality of life and in doing so I hope we can make some major breakthroughs for not only AIP patients but all porphyria patients. It is important that all porphyria patients rally around one another. Kindness doesn't cost a thing and we are all in this together!

MADELYN MAKES A DIFFERENCE

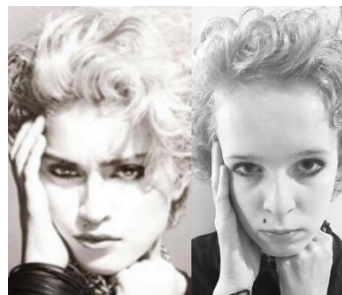
My name is Madelyn Havard and I was diagnosed with EPP when I was 4 years old. I learned how to live with it, and just thought of it as something that I would always have. Then, two years ago, I started having stomach pains and discomfort. My doctor ordered an ultrasound and it showed that my liver and spleen were enlarged. After a liver biopsy, we learned that I had cirrhosis of the liver. The damage was caused by my EPP, but my liver was still functioning properly. My specialist at UAB recommended a bone marrow transplant to cure my EPP and stop my liver damage. This did mean that I would not have EPP anymore, but there were a lot of risks involved. I live in Memphis, Tennessee, so I was accept-

ed into St. Jude Children's Research Hospital. To have a bone marrow transplant, we needed to find a donor match and I needed to have a lot of chemotherapy to kill all of my bone marrow. I was admitted on June 18, 2018, and I had my transplant on June 28, 2018. The nine days in between these two dates, I had eleven rounds of chemotherapy. All of the chemotherapy made me lose my hair. I was really sad about losing my hair. My friends and I realized once I was bald that I would make the best Eleven (from *Stranger Things*) for Halloween. I had so much fun dressing up that I decided to dress up as other characters as my hair started growing back out. I chose to put it on Instagram so that other people who lost their hair could see it and have fun with their new hair. I have been able to connect with other people who

have gone through bone marrow transplant or lost their hair, which I am really happy about. It has been about a year since I had my transplant and I am officially EPP free!

(Editor's Note: Bone Marrow Transplantation is a serious and difficult procedure. Though curative for EPP, it is not considered unless serious complications are present.)

Pictured from left: Madelyn as Will Ferrell in Elf, Ellen Degeneres, and Madonna.



PATIENT EDUCATION AND SUPPORT MEETINGS

The APF continue to host Patient Education and Support Meetings across the US. We recently hosted a meeting at Mount Sinai, Camp Sundown (NY) and Winston Salem, NC. Special thanks all of our patient members who attended and to Dr. Balwani and her team from Mount Sinai, Dr. Bonkovsky and his team from Wake Forest Baptist University in Winston Salem and the team at Camp Sundown for their support. As we approach the last quarter of the year, we are eager to schedule more meetings in a city near you. If you are interested in hosting a patient education and support meeting, please contact the APF office for more information. (Photo L-R: Kristen Wheeden, Luca Fierro, Dr. Manisha Balwani, Karli Hedstrom, Chanan Stuauffer and Dr. Hetanshi Naik) **Next stop: Boston, MA (November 2019) + Orlando, FL (December 2019)**



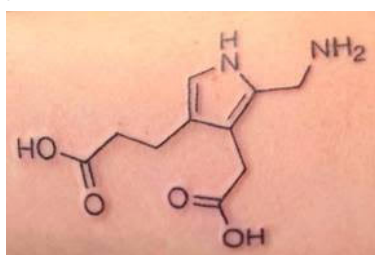
DIGESTIVE DISEASE WEEK



The American Porphyria Foundation exhibited at the 50th Annual Digestive Disease Week Conference in San Diego, CA in May 2019. We had the opportunity to meet hundreds of physicians and educate them about Porphyria. We would like to give a special thanks to Mary Schloetter for managing the booth and to Stephanie Purtrell Andrews and Beverly Purtrell for volunteering their time to help educate physicians at the exhibit. The next conference will be held next year in Chicago, IL. If you are interested in volunteering for this event and are local to the Chicago area, please contact Edrin Williams, Director of Patient Services to express your interest. You can find out more information on their website at www.ddw.org/home.

TATTOOS AND ACUTE PORPHYRIA GUIDANCE

According to Dr. Bruce Wang, Porphyria Expert, tattoo ink can get absorbed systemically and has been found in the immune cells of the liver (macrophages) one year after tattoo in mice that were tested. However, there is no data on whether the ink is metabolized and there's no evidence of liver damage. It should be safe in AHP patients.



Tattoo pictured belongs to Rebekah Gidley.

PANHEMATIN HISTORY



APF member, Sandra Ihrig (Culver, OR) came across this story during Women's History Month. Marion J. Finkle, M.D. (pictured left) was head of Orphan Products at the FDA when Panhematin was approved in 1983. In the mid-1970s, she chaired the agency's Committee on Drugs of Limited Commercial Value. She ushered in FDA's administration of the 1983 Orphan Drug Act, which aimed to provide medicines for rare, commercially untenable diseases through a variety of inducements to industry. She left the agency in 1985, by which time her office had designated 54 drugs with orphan status, 31 of which were either on the market or well on their way. The first orphan drug was Panhematin – and Desiree Lyon worked with Dr. Finkle to get it approved! <https://www.fda.gov/AboutFDA/History/VirtualHistory/HistoryExhibits/ucm346388.htm>



David Yates, CEO, CMA and Sian)

A SPECIAL DAY FOR SIAN!

“Wow, oh wow!” was Kimberly Dawn Merrill’s reaction when she and her daughter, Sian, heard that they had been selected for an amazing opportunity at Clearwater Marine Aquarium in Clearwater, Florida. The Inspire Program at this aquarium hosts people who have been especially inspired by Winter the dolphin through trials of their own experience for a special VIP experience, free of charge. This was an amazing opportunity for Sian, who lives with EPP. Sian and her family used the opportunity raise awareness about EPP!

GLOBAL GENES 2019 PATIENT ADVOCACY SUMMIT

Global Genes will host their annual Patient Advocacy Summit in San Diego, CA on September 18-20, 2019. This annual event brings together rare disease advocates, industry leaders, patients, healthcare partners and many others who care about rare! Global Genes provided a travel scholarship to enable people within the rare disease community to participate in the annual conference. Amy Chapman, Director of Social Media will be joined by a host of patients who received the scholarship to attend this event. We can't wait to hear about their experience while at Global Genes. If you are local to the area, there is still time to register for this event. Please visit the Global Genes website for additional information on registration fees, etc. www.globalgenes.org.



ICPP IN MILAN, ITALY

The APF will be attending the International Conference on Porphyrins and Porphyrins in September. We will prepare two abstracts, moderate multiple sessions, and present on Patient Day. The scientific conference will be an opportunity to learn the latest in porphyria research and bring it home to share with you! Most important, our expert doctors of the Porphyrins Consortium and several Protect the Future physicians will have the opportunity to lead sessions and collaborate with colleagues worldwide.

HAMZA LAB AT THE UNIVERSITY OF MARYLAND (UMD)



HAMZA LAB

The Hamza Lab at the University of Maryland is led by Principle Investigator and researcher, Dr. Iqbal Hamza. The critical work done in this lab explores the transport of heme. Porphyria is a group of eight diseases that are all based in the development of heme. Consequently, the more that is understood about how heme operates, the greater hope we have for future treatments. **“Working at the interface of biochemistry, cell biology, and molecular genetics on fundamental questions related to iron and heme trafficking with implications in human health and disease,”** is the mission of this lab. This is yet another way that we support research – the key to your cure! For more information on the Hamza Lab, visit <https://hamza.umd.edu>.



This summer, Dr. Hamza provided access to a young learner who was able to gain experience in a lab and to learn about the function of heme. Zach Wheeden shadowed Xiaojing Yuan, Ph. D, (far left in photo). It was a valuable experience and enabled him to learn porphyria at a basic science level.

Photo: Dr. Hamza (center back), lab staff, and student, Zach Wheeden (second from left).



TIPS FOR YOUR DOCTOR'S VISIT

Have you ever arrived at a doctor appointment feeling unprepared or left an office without getting the answers you wanted? Make the most out of each appointment with your physician by following this important advice.

Make A List: You may not always need to share all your information with every doctor you see but the following items are particularly important: A list of all your medications and needed refills, a summary of your medical history, a list of your recent tests, a list of your questions, concerns and new information, forms your doctor needs to address,

Plan Ahead for Your Doctor Visit: Prepare your questions in advance limit them to three questions for each visit and a list of your symptoms (For example, racing heart, blisters, etc.) Be concise. When you schedule your appointment, ask if you should have test results or other medical records sent to the doctor's office before your visit. Nothing is worse than rescheduling for new

tests you could have taken earlier or not had with you.

At Your Visit: Be on time. Give and expect respect. Bring your list and tell the doctor what you want to discuss and your goals for the visit. Be as brief as possible. Communication is an especially important skill. Make every word count because the doctor may only have 15 minutes to spend with you.

Be sure you understand what the doctor is advising you. If not, ask questions until you understand. If there is not enough time for all your questions, ask for handouts and brochures that will give you more information or schedule another visit.

You and your doctor may have different goals for the visit. For example, your doctor may want to just check your blood pressure, while you may have worries about possible surgery.

Many things can get in the way of helpful communication; *emotions, communication style, different goals and lack of time all work against us.* When emotions are high,

logic is low. If you find that your emotions are interfering with your visit, explain this to your doctor. Try taking a moment to reflect on what you want to say and try again.

Lastly, you may feel that you know more about certain aspects of porphyria than your physician. Major medical journal articles are usually best accepted than internet articles. Please contact the American Porphyria Foundation by calling 1-866-APF-3635 or visit our website.

After the Visit: Often patients have questions they forgot to ask. If it is urgent, call the office right away. Otherwise, check the Patient portal which is free and often have a mobile app to download. Medical Staff usually look and try to answer the same day. If you still don't have the answer, call your doctor. However, it is best to have the question clearly written. Be aware that the doctor may not be able to answer your call until 24-48 hours, but a nurse or physician's assistant may be able to help earlier.

The American Porphyria Foundation has a wonderful toolkit available at www.porphyrifoundation.org.

IN MEMORY & IN HONOR



We thank the families and friends who memorialized their loved ones with a generous gift to the APF.

IN MEMORY:

Carole Kuklewski for **Vince Kuklewski**.

We also thank those who honored a friend or family with a generous donation to the APF.

IN HONOR:

The Lewis Family for **Joe and Ben**; Teresa Heedles for **Morgan McKillop**; Kerri Oman for **Jonathan Pultz**; Blueport Blue Point UFSD for **Morgan McKillop**; Thomasina Taylor for **Nathan Carr**; Andrew Blakesless for **Alex Blakeslee**; Phyllis Liddell for **Paul Kraft**.

FAREWELL, AUTUMNLEE!

AutumnLee Meyer has spent the last year with the APF as Administrative Assistant. She will be moving on to focus on her studies to become a counselor. We wish AutumnLee the best of luck in her future endeavors and many thanks for her hard work at the APF!

The information contained on the American Porphyrria Foundation (APF) Web site or in the APF newsletter is provided for your general information only.

The APF does not give medical advice or engage in the practice of medicine. The APF under no circumstances recommends particular treatments for specific individuals, and in all cases recommends that you consult your physician or local treatment center before pursuing any course of treatment.

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VOLUNTEERS NEEDED!

The American Porphyrria Foundation will be exhibiting at the Annual Liver Meeting (AASLD) in November and the Annual Hematology Meeting (ASH) in December. We are looking for local patients who are interested in volunteering at the booth at either event.

These events will give you the opportunity to share your story and to help educate physicians who visit our booth! Multiple time slots are available to accommodate your schedule.

If you are local to the Boston or Orlando area and interested in volunteering at these events, please contact Edrin Williams, Director of Patient Services via email at edrinw@porphyriafoundation.org or via phone at 301.347.7166 for more information.



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