



AMERICAN PORPHYRIA FOUNDATION

2nd Quarter, 2019

★★ 2019 ★★  
**PRESIDENT'S AWARD**  
RECIPIENTS

We are proud to announce the recipients of the American Porphyria Foundation President's Award 2019. The President's award is a time-honored tradition of the APF to honor members and partners who have gone above and beyond to support the foundation or our members, and who have increased awareness of porphyria. Congratulations to our 2019 awardees: **KARL ANDERSON, MD, AMY BURKE, PAUL STICKLER and TERRI WITTER.**



**KARL ANDERSON, MD** Dr. Karl Anderson, a physician at the University of Texas Medical Branch (UTMB) at Galveston, Texas, is considered one of the top porphyria experts in the world and has served on the American Porphyria Foundation Scientific Advisory Board (SAB) since its inception in 1982. He never fails to go above and beyond in his support of the APF and his care for his patients. We are blessed to have Dr. Anderson as a clinician and researcher!



**AMY BURKE (MEMBER)** Amy was diagnosed with Porphyria Cutanea Tarda (PCT) in 2009 and has since been an inspirational member of the APF. She is a strong advocate for porphyria awareness and a support among her peers, serving as an administrator on our Facebook support groups. Amy is always working behind the scenes! The APF can always count on Amy to lend a hand when needed. We are lucky to have her as an advocate and volunteer.



**PAUL STICKLER** Paul is the Vice President of Commercial Operations at Recordati Rare Diseases and is a dedicated friend to the APF and to the porphyria community. Having worked in rare disease for 13 years, he is deeply committed to helping patients. His commitment has helped acute patients gain access to life-saving Panhematin® treatment. We are proud to

work with Paul, and this award recognizes his hard work for our members and support of our mission.



**TERRI WITTER (MEMBER)** Since her diagnosis with Acute Intermittent Porphyria 41 years ago, Terri has been a fierce advocate for porphyria research, awareness, and peer support. She has been involved in seven research trials, educated medical students and nursing staff, and presented to the FDA. Terri is an active member of APF social media and is always there with encouragement,

comfort, and something to make you smile.



**YVETTE STRANGE, 25 YEARS OF SERVICE TO APF**

Yvette has been a beloved and valued member of the APF team since 1994. We have been blessed to have her hard-working and quiet energy. With the office move to Bethesda, Yvette remained in Houston working on special projects for the APF. "In 1994, the year I was hired, one of the first jobs I did was to type a newsletter. Desiree wrote most of the newsletter longhand, which I then typed on a small laptop computer. We didn't have a mail service to send out the 1,000 newsletters, which had to be manually sorted and delivered in trays to the Post Office. When email became popular, we were able to communicate and answer patient questions on a more-timely basis and members could support each other by sharing their concerns and experiences. I have numerous fond memories with Desiree and the staff. I'm very proud I have had the opportunity to work with the APF, a prestigious organization of people working diligently together to help ease the pain of so many porphyria patients." **Thank you for 25 wonderful years, Yvette!**

 **RECORDATI RECORDATI NEWS**

Orphan-Europe has been the European arm of Recordati Rare Diseases since 1990, serving 30 countries, with access to Panhematin® and Normasang. With nine affiliates, Orphan-Europe spread through Europe, the Middle East, North Africa, Central and Eastern Europe, Russia, Asia-Pacific, Latin America, the USA and Canada. Recently, Orphan-Europe underwent a name change and will now share the name of their counterpart, Recordati Rare Diseases. The APF has collaborated with Recordati since the beginning, and will continue our partnership across the globe. Panhematin® has saved thousands of patients and now is being used widely to prevent attacks.



### SCENESSE FDA UPDATE



The FDA granted Scenesse a PDUFA (Prescription Drug User Fee Act) date of July 8, 2019, under Priority Review status. By that date, the treatment will either be granted market authorization or be given a Complete Response Letter (CRL) to communicate that the review is complete and the treatment has not been approved for marketing in its present form. Clinuvel, the sponsor, has been in frequent contact with the reviewers at the FDA to respond to any inquiries. The APF has continued advocacy efforts to work toward approval of this life-altering treatment!

### EPP CLINICAL TRIAL - ENROLLMENT COMPLETE!



Mitsubishi Tanabe Pharma We are delighted to share that the clinical trial for MT-7117 has reached full enrollment of 102 participants. This clinical trial filled within 10 months. We appreciate all the hard work and dedication that each of you put into filling this clinical trial. Whether you volunteered, shared with your family and friends, or supported participants - you are all medical heroes.

A special thank you to our physicians and research coordinators for their hard work and dedication to this clinical trial.



### ALNYLAM® PHASE 3 RESULTS

APF Scientific Advisory Board member and porphyria expert Dr. Manisha Balwani presented the complete results of the Phase 3 study of Givosiran at the European Association for the Study of the Liver (EASL) in April. Givosiran is an investigational RNAi therapeutic in development for the treatment of acute hepatic porphyria (AHP). The results showed that Givosiran met the primary efficacy endpoint and many of the secondary endpoints.

### TWO TRUTHS AND A LIE | Christina Verkest



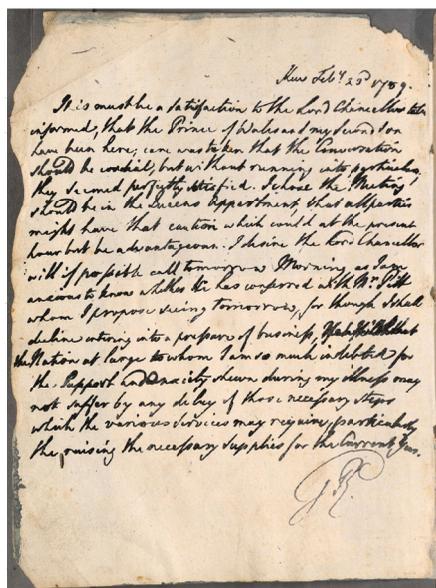
I am a 27-year-old from the suburbs of Detroit. I was diagnosed with EPP when I was about 5 or 6 at Children’s Hospital in Downtown Detroit. Since I have dealt with my EPP for the majority of my life, I am very comfortable with it, and even when I don’t feel as “fashionable” in the summer sun, I know how to own it and

am happy to educate others about why I might be wearing long pants and sleeves in the middle of a sweltering July day. So I try to this bring this “own it, even the not-so-great stuff” attitude into my high school Spanish classroom, as almost all of them can relate in some way. I always spend time at the start of the school year ensuring that my classroom environment is one that allows students to feel comfortable, accepted and supported. So one of the “ice breakers” I like doing is the infamous “2 truths and 1 lie,” where I share with my students two truths about my life and

one lie. They have to guess which of the three is my lie. I like to get really obscure with these because it makes an impression on the kids, but it also allows me to be real with them about who I really am.



One of my truths is “I am a modern-day vampire.” That usually catches their attention! After many of them guess that I am lying about being a vampire, I tell them that, in fact, I am! I take the time to explain that I have a rare genetic disorder which causes me to experience photosensitivity or pain when I am in the sun. I even paint them the picture of my “space suit,” which refers to the long pants, sleeves, hat and sometimes gloves that I have to wear when I go to amusement parks or the beach. While sharing this information at such an early point in the school year may require me to get somewhat vulnerable with the students, my hope is that I can educate them about the disease but also spread some awareness about the unique characteristics every one of us has.



### KING GEORGE LETTER

A fascinating artifact – which was made available to the American Porphyria Foundation and is available on the APF website – is this handwritten and signed letter from King George III, the only known document to mention the illness that labeled him “Mad King George.”

#### TRANSLATION:

*It [is] must be a satisfaction to the Lord Chancellor to be informed that the Prince of Wales and my Second son have been here; care was taken that the conversation should be cordial, but without running into particulars, they seemd perfectly satisfied. I chose the Meeting should be in the Queens apartment, that all parties might have that caution which could at the present hour but be advantageous. I desire the Lord Chancellor will if possible call tomorrow morning as I am anxious to know whether he has conferred with Mr. Pitt whom I propose seeing tomorrow, for though I shall decline entering into a pressure of business, [...] /yet I wish ?/ the Nation at large to whom I am so indebted for the support and anxiety shewn during my illness may not suffer by any delay of those necessary steps which the varioius services may require particularly the raising the necessary supplies for the current year. GR*

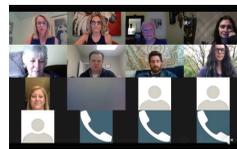


# PORPHYRIA AWARENESS WEEK 2019

Porphyria Awareness Week was a success! Thanks to our amazing APF members and partners. PAW is an annual opportunity to create awareness among physicians, in your community, and in the general public. The theme of the week was "Paint the World Purple" as porphyria is named from the ancient Greek word *porphyros*, meaning purple. The week was full of events big and small, social media outreach, and collaboration with our industry partners. Thank you to everyone who participated!!

## Events

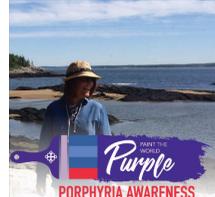
**JASON SUMNEY** held a vintage mixer to raise awareness \* **THE MCKILLOP FAMILY** held a Fun Run from the Sun fundraiser with 400 attendees! \* **MURPHY MCNUTT** held a fitness class to sweat for porphyria – raising \$400! \* **EDEN LIGHTHALL** held a lemonade stand to raise funds and awareness! \* **HATTIE STUHLSTAZ** read *Shadow Jumpers* with her class to explain EPP \* Nicole Castellano held a *Let's Talk Porphyria* video conference \* **THE ORPHAN DRUG CONGRESS** shared our theme for 1,000 doctors and industry reps to see \* **THE COMMUNITY WORE PURPLE FOR PORPHYRIA!!!**



Photos (left to right): Jason Sumney (AIP); Morgan McKillop (EPP, age 9); Eden Lighthall (age 9, daughter of Brandy Lighthall); Murphy McNutt and group (PCT); APF staff (Bethesda); Hattie Stuhlsatz with *Shadow Jumpers* book; Bryce Payne; Amy Burke; center stage at Orphan Drug Congress; ZOOM video conference.

## Media

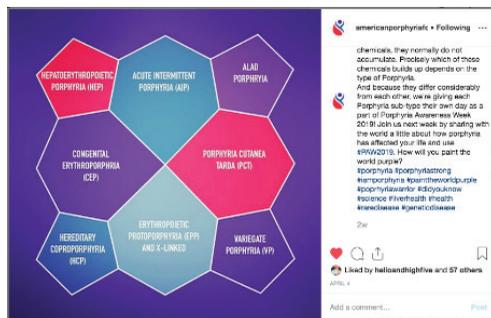
**FACEBOOK \* TWITTER \* INSTAGRAM** Daily facts, member stories, and even a trivia contest was held on social media. Many members held APF fundraisers – and we are grateful for the impact you made. #APFpainttheworldpurple #APFporphyriaawareness #PAW2019 #porphyriastrong



clairebearian Posted about it on social media and shared my story on the "Let's talk Porphyria" webinar!

jazreel954 I posted the campaign on FB and educated my nurses in the infusion center at Emory University Hospital

angelmyheart52 Wore my purple and posted pictures, held an 8 day fundraiser and raised over \$1000 and participated in the Let's Talk Porphyria Thursday evening!



## Trivia

**WINNERS:**  
 Rebekah Gidley - open group;  
 Abdul Butt - CEP;  
 Stacie May Cromack- PCT;  
 Tara Cantley - AIP;  
 Bekah Lock - EPP

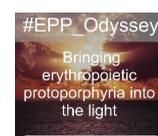


## Pharmaceutical Partners

- ALNYLAM PHARMACEUTICALS** implemented targeted advertising information about porphyria diagnosis that was picked up across the U.S. They also ran internal awareness events.
- Houston:** <https://www.chron.com/sponsoredarticles/lifestyle/women/article/How-to-Pinpoint-a-Rare-Disease-That-Can-Often-be-13750423.php>
- Albany:** <https://www.timesunion.com/sponsoredarticles/lifestyle/women/article/How-to-Pinpoint-a-Rare-Disease-That-Can-Often-be-13750423.php>
- Siskiyou:** <https://www.siskiyoudaily.com/ZZ/sponsored/20190407/how-to-pinpoint-rare-disease-that-can-often-be-misdiagnosed-for-up-to-15-years>
- Canton:** <https://www.cantonrep.com/ZZ/sponsored/20190407/how-to-pinpoint-rare-disease-that-can-often-be-misdiagnosed-for-up-to-15-years>



**RECORDATI RARE DISEASES** held a click campaign to drive awareness through the APF website. For every click up to 1,000 clicks, Recordati donated \$5 to the APF!



**CLINUVEL** ran a series of awareness-raising posts, centred around the diagnostic odyssey of patients with EPP.

## CEP INITIATIVE – EDUCATION AND SUPPORT | Amy Chapman, APF



A year or so ago, I met a wonderful CEP Patient, Abdul Butt from Pakistan, through the APF Facebook Group. Listening to him tell his personal journey about the rarest type of cutaneous porphyria (CEP) made me want to learn more and to help people with CEP gain APF assistance. My new friend found that the APF Facebook group was a safe place for guidance and help to receive a proper diagnosis. Through many calls and questions, I came to understand how they endure living with CEP in a country with little to no support. Abdul became an Ambassador for the APF in Pakistan and together we found about 10 additional CEP

patients. There is a genuine need to help our new-found friends. Listening to their medical needs, we learned of the difficulties finding a physician willing to help patients. Thus, getting a proper diagnosis in Pakistan is nearly impossible. These patients must completely avoid sunlight in a country that is sunny and has highly dangerous living conditions. Acquiring simple things, such as protective clothing and sometimes even food, can be difficult in Pakistan. The most horrific thing I learned about was the lack of hospital care and concern. They are treated as though they do not exist, even being told that they are a curse to their communities and families. What a discouragement! I had the opportunity to speak over the phone with one doctor in Pakistan – it was a nightmare. When we had our next Ambassador meeting, I explained and even apologized to them. I was ashamed. They

said, “You see, Amy, we have to deal with this every time we go to the hospital or doctor.” Enough of that, I said! With the help of the Porphyrrias Consortium, the APF and our Pakistan Ambassadors, we have had a great response from patients in Pakistan. The APF will be providing DNA testing for those who are symptomatic.

We hope to find physicians in Pakistan who are willing to learn about CEP through our comprehensive Physician Education Kits. We hope someday to have an expert physician train them and assist when they have a CEP patient. Education also must include our CEP patients – teaching and sharing with one another no matter where we live. We can thrive as a rare group. Never be silent ... you have a VOICE!

(Photo: Flowers sent to Abdul in appreciation for his work as an Ambassador)

### CEP INTERNATIONAL SUPPORT CALL

The APF hosted a virtual Patient Education & Support meeting in March for our international CEP members. This meeting brought our CEP members together in a safe environment to learn about their disease and listen to a presentation by porphyria specialist Dr. Angelika Erwin, Cleveland Clinic, who also answered patient questions related to diagnosis, treatment and management. For many, this was the first opportunity that they were given to speak directly with a physician and to engage with other members of the community. We look forward to having another CEP Webinar with our remaining USA members.

### PATIENT EDUCATION MEETINGS – COMING TO A CITY NEAR YOU!

The American Porphyria Foundation has hosted nine Patient Education and Support Meetings. With the help of our community, each meeting has been a success. The meetings give patients and families the opportunity to meet others with similar experiences. These events are attended by a porphyria expert, in person or via Skype. **NEXT STOP:** May 31, Salt Lake City, UT | Santa Rosa Beach, FL | Birmingham, AL | Boston, MA | Orlando, FL | Winston Salem, NC | Chicago, IL | New York, NY

**If you are interested in hosting a Patient Education and Support Meeting, please contact Edrin Williams, Director of Patient Services, at 1.866.APF.3635 or via email at [edrinw@porphyriafoundation.org](mailto:edrinw@porphyriafoundation.org).**

### PORPHYRIA SPECIALISTS AT RECENT MEETINGS



Amy Dickey, MD (left)  
BOSTON, MA



Manisha Balwani, MD  
SEATTLE, WA



Samuel Silver, MD  
ANN ARBOR, MI



Danielle Nance, MD  
PHOENIX, AZ

### PROBIOTIC AND PREBIOTIC

Probiotics are very popular, and our current understanding of them is quite limited. A probiotic is a mixture of a few strains of normal gut bacteria that have been deemed “good” and are already in our diet, especially in fermented foods like yogurt or sauerkraut. There is no acute porphyria concern. The role of prebiotics, in theory, is to boost the growth of the bacteria in the probiotic supplement and amplify their good effects. PREbiotics are supposed to be digestible only by bacteria, not the human host. Many are large sugar molecules, which may cause gassy discomfort in humans. This could be a problem in AIP, where intestinal motility is impaired as part of the disease. Also, gassy discomfort could be mistaken for a porphyria

attack. The evidence that current supplements are beneficial is quite limited. A PRObiotic from a known reliable source is harmless even if not as useful as the seller suggests. Yogurt is cheaper and tastes better. I advise against PREbiotics. Also, pro- and pre-biotics are considered “food,” which means little or no FDA oversight of manufacturing standards and purity. Chinese herbs at one time were being adulterated with prescription drugs (such as steroids and antibiotics), and the same may be going on with prebiotics. Most people see supplements as doing no harm and possibly some good. However, in the case of pro- and pre-biotics in acute porphyria, it is unclear. (D. Montgomery Bissell, MD, Director, UCSF Porphyria Center, University of California)

## RESEARCH. IF NOT YOU, THEN WHO?

If you have ever asked a question about your type of porphyria only to have a physician respond with “I don’t know,” you are a candidate to participate in research. The American Porphyria Foundation is actively recruiting research volunteers for all types of porphyria. We are calling on our patient members to boost the number of volunteers that participate in these studies. The government grants that fund rare disease research, through the National Institutes of Health, support a limited number of disease groups like porphyria. A critical area for assessment to fund a rare disease group is the number of participants. If there is not proof of steady growth in the number of research volunteers, we risk losing critical funding that supports our Porphyria Centers across the US. To secure ongoing funding, we urge you to participate. It is easier than you may think! We understand that it is an important and personal decision, but if not you ...then who??? If you are interested in volunteering for any of these studies, please contact Edrin Williams, Director of Patient Services, at [edrinw@porphyriafoundation.org](mailto:edrinw@porphyriafoundation.org) for additional information.

### Some available opportunities:

- » Longitudinal Study (All Porphyrias)
- » Harvoni Study (PCT)
- » Panhematin® Prevention Study (AIP, VP and HCP)



REMEMBER ...

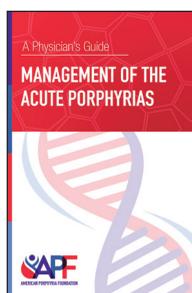
**RESEARCH IS THE KEY TO YOUR CURE**

## INTER-AGENCY TASK FORCE ON PAIN

**PAIN**

The third meeting of the Pain Management Best Practices Inter-Agency Task Force took place on May 9 and 10 in Washington, DC. The Task Force voted on the final draft of the Task Force recommendations for acute and chronic pain management. **THANK YOU to all APF members who took the opportunity to submit comments on this important issue. You took the tremendous opportunity to have your say about the need for safe opioid pain management and individualized care for acute porphyria.**

## NEW APF BROCHURES!



We are excited to share our updated brochures. Members of the APF Scientific Advisory Board reviewed and updated content for our new brochures which include *An Overview of the Porphyrias*, *Management of the Acute Porphyrias*, *A Guide to Panhematin*, *A Guide to Porphyria Cutanea Tarda*, and *A Guide to Erythropoietic Protoporphyrinuria*. This valuable information will be included in patient and physician education packets.

## WORDS MATTER by Sharon Dill



I’ve been thinking a lot about words and language. When we travel or move to different places our words often change, sometimes our language completely changes. I lived in Alabama for the first part of eighth grade and quickly, without realizing it, began to adapt my cousins’ Southern drawl. When I moved back to Massachusetts, my Yankee speech pattern took over again. For my 40th birthday I traveled to Italy and spent 10

wonderful days in the Tuscany area. Along the way I tried to pick up a few Italian words to make communication a bit easier. In June 2014 I made the biggest move of my life, yet I realize that after almost five years, my words and language haven’t changed. I’ve learned a few new words, but the words that I use, the meaning and language behind them, have stayed the same. They haven’t changed. My move was not voluntary, I resented it, and frequently hoped it was just a horrible nightmare that I would wake up from. But the move was real, and it is permanent. The move was to my new reality of Variegated Porphyria. My world, my life, has changed just as much, perhaps even more so than if I had moved to a far-off foreign country. Yet I’m still using the same words and attaching the same meaning as in my old reality, my old life, especially the words I tell myself, the self-talk, the self-critiques. “I didn’t even get my bed made,” “another day I got nothing accomplished,” “I only got half the things on my to-do list done,” “I didn’t get supper made – again.” These are the words I hear constantly replaying in my head, criticisms for what I haven’t done – nothing about the things I did get done. But I’m starting to realize that these are the words and language spoken where I used to live, and maybe it’s time to start learning the words and language of where I live now – in the world and reality of a strange, weird, complex medical condition that is both chronic and acute. In this new place I live, I need to change the words and language for the messages I tell myself. Instead of the criticisms for what I didn’t get done, I need to recognize what I did get done. “I did make it out of bed and into the TV room,” “today I rested and gave my body what it needs to fight,” “even though I felt lousy today, I got several things accomplished.” Learning this new language isn’t easy, there isn’t an app I can download on my iPhone to help with the translations. It will take time and practice. But maybe there are some rewards to help me along the way: less self-loathing, more self-respect, more self-confidence as I slowly begin to learn and embrace the words and language of living with Variegated Porphyria.

## UNITING ART AND SCIENCE



Stay tuned for the big reveal of an APF member’s portrait which will be exhibited at a major hospital this fall. *Beyond the Diagnosis* commissions artists who donate their time and talent to paint orphan disease patients for this traveling exhibit. Their goal is to unite art and science to inspire research and innovation for children living with a rare disease.



## ACMG CONFERENCE

The APF hosted a booth at the American Clinical Medical Geneticists conference in March in Seattle, WA. The opportunity enabled us to educate many physicians on porphyria. Special thank you to member Tirzah Idahosa, who came for an entire day to help! We also were visited by our friends from Brazil (Dr. Charles Lourenco, who trained under Dr. Karl Anderson at UTMB, and students, pictured left). **Next up: DIGESTIVE DISEASE WEEK, San Diego, CA, May 18-21**

## THE PURPLE STORE

This Seattle-based store seems the perfect place to raise awareness about porphyria. Owner Adam Sheridan shared that customers often comment that their urine might turn purple if they eat the syrup, jam, or honey on display. “Don’t joke,” replies Adam, “there is a serious disease called porphyria ...” Even The Purple Store can help our effort!



## RARE PATIENT ADVOCACY SUMMIT



This year the Global Genes annual RARE Patient Advocacy Summit will take place in San Diego at the Sheraton San Diego Hotel & Marina. We would love to have the porphyria community represented. This event brings together hundreds of patients, patient advocates, industry leaders and more. If you register before July 31, 2019, you will get the discounted Patient/Patient Advocate rate of \$175. Registration to this event includes a two-day agenda with Ted-style keynotes, five tracks of educational content, access to workshops, exhibits, meals, networking activities and the RARE Champion of Hope Celebration. Please register using the link below. The APF is not responsible for any travel or accommodations. <https://globalgenes.org/event/patient-summit/>



## NEW MERCHANDISE IN THE APF STORE!

T-SHIRTS, UMBRELLAS, PRINTED SAFE / UNSAFE DRUG LIST, AND MORE!



THE UNIVERSITY OF UTAH

## ONE IN A MILLION

John Phillips, PhD, esteemed member of the Porphyrias Consortium and researcher at the University of Utah, shared a recent initiative that profoundly affected the life of one patient, Tyler. *One In a Million* is a must-see short film about the work of the U of U researchers and physicians. “This remarkable documentary beautifully portrays Tyler and his family’s experience living with disease, and their journey to hope with University of Utah Health,” said Michael L. Good, MD, CEO, University of Utah Health. “The film is a proud testament to the heroic, life-changing work that goes on here every day. We are excited to help share these stories with the world.” Stories like Tyler’s would not be possible without research and advances in science and medicine. You can see the film here: <https://www.youtube.com/watch?v=WlupO5-F-28>



## A FACEBOOK BIRTHDAY GIFT!

Kim Jacobs was diagnosed with AIP about 10 years ago; however, this last year was especially difficult. Kim spent almost as much time in the hospital as she did at home. Kim has the amazing support of her daughter, Krystal. This year, instead of birthday gifts, Krystal raised funds for the APF through Facebook. Krystal received a message from Facebook that said if she would like to set up a donation page, they would donate the first dollar. Krystal chose the APF as her charitable organization and set her goal at \$200, which she accomplished on her birthday! Everyone in the Jacobs’ household knows what to do and how to handle an emergency with Kim since they see it all the time. They hold a cure – or anything that helps – close to their hearts. Krystal graduated last year and is planning to become a police officer. (Photo: Kim Jacobs and daughter, Krystal)



**THANK YOU, KRYSTAL, AND THE MANY OTHERS WHO HAVE DONATED TO THE APF THROUGH FACEBOOK!!**



## “TIC TAC TOE”

Did you know that acute porphyria is often known as the “tic tac toe” disease? The scars from the many surgeries and procedures, from diagnosis through treatment, create the look of a tic tac toe board on a patient. Candace Johnson recently went through additional procedures and bravely showed her scars on social media. Candace shared, “I wear my scars with pride and a reminder that others are going through much worse.”



## “IRON HACK”

The National Center for Biotechnology Information (NCBI) hosted an “Iron Hackathon” February 25-27, 2019. During a Hackathon, teams spend between 24-48 hours building, creating and delivering a product. The idea is to collaboratively code in an extreme manner, to start from scratch and end with a working prototype. This hackathon focused on the genomics of Iron-linked Rare Diseases. Teams spent three days addressing a challenging set of scientific problems related to a group of datasets, analyzing and combining datasets to work on these problems. Porphyrias Consortium members Dr. Robert Desnick and Dr. John Phillips presented on genomics and iron metabolism. Kristen Wheeden, APF, spoke on the patient perspective and the *Protect the Future* initiative.

# ICPP

Milan, 8-11 September 2019

International Congress on Porphyrins and Porphyrins  
FROM BENCH TO CARE



**International Patient Day will be held on Saturday, September 8th, in Milan, Italy. Registration is currently open.**



The McKillop Family:  
Ben, Colleen, Kate (13),  
Tyler (11), and Morgan (9)

## FIND YOUR SHADOW 2019: CONGRATULATIONS TO THE MCKILLOP FAMILY!

A main goal of Shadow Jumpers is to help kids living with Erythropoietic Protoporphria to do something they have always wanted to do but have not been able because of the sun. Find Your Shadow 2019 is sending one such family to Disney! We received many wonderful submissions and we wish we could select them all. Find Your Shadow 2019 congratulates the McKillop Family! Morgan McKillop, age 9, lives with EPP. She and her family are headed to Disney this summer! APF Shadow Jumpers program, through private donations and volunteers, will plan a safe trip for Morgan in spite of the sun!



### Q & A WITH PORPHYRIA EXPERT, DR. BRUCE WANG, UCSF

The APF asked our Facebook friends for their top questions they would ask a porphyria expert. The following questions were submitted to Dr. Wang for his responses ...

#### **Q. Does EPP give us bad teeth? Also, do people with EPP get stomach pains or is that with the other porphyrias?**

A. The porphyrin that accumulates in EPP patients is protoporphyrin IX, which does not cause discoloration to teeth or abdominal pain. The type of porphyria that leads to discolored teeth is Congenital Erythropoietic Protoporphria. The porphyrias that lead to episodic abdominal pain attacks are the acute hepatic porphyrias.

#### **Q. I have EPP and I have a severe reaction on my hands and lips. Do I seek urgent care? Also, what can you even do when you burn your lips?**

A. The acute reactions to sunlight in EPP can be very severe and,

unfortunately, there are not many effective options to treat the symptoms. Nonsteroidal anti-inflammatory drugs (NSAIDS such as ibuprofen) and stronger pain medications like opiates often provide only partial relief. These symptoms generally resolve spontaneously within hours to days.

#### **Q. Do children under 10 years of age really have AIP attacks? I have seen this on social media, and to my knowledge it starts in teenage years with hormones.**

A. Acute porphyria attacks occur when the body, and in particular the liver, has increased demand to make heme that the mutated enzyme cannot keep up with. Hormones are known to be important inducers of heme production by the liver. This is why more than 90% of the patients who have acute porphyria attacks are female, and also why the vast majority of the attacks occur during the age range when they are actively menstruating. But there are other causes that can induce the liver to make more heme, so it is possible, though rare, for acute attacks to occur prior to the start of menses. I would recommend carefully looking for other causes of acute, episodic abdominal pain in younger kids in order to not miss other more common causes.

## IN MEMORY & IN HONOR



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

### IN MEMORY:

Kathryn Bauer for **Allen Bowden**; Charlotte Beck for **Elva Denger**; Anonymous for **Albert and Geraldine Gilmartin**; Anne Oleary for **Harry Grebowich**; Charlotte Wiedman for **Alice Ryback**; Gloria Sheehan for **Paul Sheehan**; Audrey Sokol for **Renee Lawrence Sokol**; Mr. and Mrs. Armstrong for **Rev. Thomas Beck**; Cheryl Czuba for **Rev. Thomas Beck**; Shoreline Senior Services for **Thomas Beck**; David Wilson for **Rev. Thomas Beck**.

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

### IN HONOR:

Barbra Weidlein for **Jocelyn Armes**; Charlotte Beck for **Cheryl Beck**; Bobby Bucher for **Minnie Bucher**; Crystal Warfield for **Patricia Hare Carter**; Artysmartys Vintage and Beyond for **Ralph Gray**; Eric Gray for **Ralph Gray**; Cynthia McCabe for **Nicholas Guancile**; Barbra Haskell for **Tara Haskell**; Rajeev Rawat for **JP Krause**; Roberta Migddon for **Sheila Landau**; Mordechai Erann for **Eric Lifschitz**; Sharon Koch for **The Liguori Family**; Jennifer Ewing for **Desiree Lyon**; Annah Dillard for **Gayle McGuffin**; Megan Bachmore for **Morgan McKillop**; Bayport Bluepoint Little League for **Morgan McKillop**; Richard Campisi for **Morgan McKillop**; Meghan Edden for **Morgan McKillop**; Rosemarie Gelling for **Morgan McKillop**; Kelly Gibbons for **Morgan McKillop**; Janet Kuzow for **Morgan McKillop**; Peter Manisalco for **Morgan McKillop**; Elliot Margolis for **Morgan McKillop**; Colleen McKillop for **Morgan McKillop**; Heather Morreale for **Morgan McKillop**; Nyree-Roviello-Meadows for **Morgan McKillop**; Jennifer Snyder for **Morgan McKillop**; True North Sales Inc for **Morgan McKillop**; Deborah Ventre for **Morgan McKillop**; Martha Burrell for **Diana Sabella**; Maria Gulluni for **Nora Slate**; Pittsburgh Vintage Mixer for **Jason Sumney**; Charlotte Beck for **Cheryl Sundblom**; Carole Aitchison for **Brady Wheeden**; Sharon Wheeden for **Brady Wheeden**; Tara Young for **Dan Yelen and Tracy Nudo**.

## What's New at the APF

[www.porphyrifoundation.com](http://www.porphyrifoundation.com)

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.

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### NEW OFFICE!

The APF has moved to Suite 200 in the same building. Please stop by to visit us!

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

**Our *Protect the Future*** program to train future experts is important. Please consider making a donation to this program. Yours and your children's future health depends on each of us supporting the training of doctors who will know how to treat us and perform research when our present experts retire.

**DON'T FORGET TO DONATE. YOUR HELP IS NEEDED TO EDUCATE PHYSICIANS AND PATIENTS AND SUPPORT RESEARCH-THE KEY TO YOUR CURE!!!**



AMERICAN PORPHYRIA FOUNDATION

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