



AMERICAN PORPHYRIA FOUNDATION

3rd Quarter, 2018

## PHARMACEUTICAL NEWS

### EPP PHASE 2 CLINICAL TRIAL – NEW TREATMENTS START WITH RESEARCH!



Mitsubishi Tanabe has developed a new oral treatment for EPP that is expected to increase sunlight duration and tolerance in individuals with EPP. Research is now underway with a Phase 2 clinical trial to investigate how it works. What is the most important element in research? YOU! You now have an opportunity to participate in this trial. YOU are our hope for the future. The study duration is 22 weeks including follow-up. You must be age 18-70 to participate and have a confirmed diagnosis of EPP. All travel is included and will be arranged by a concierge service. You will need to record your daily exposure in an e-diary – and you will never be expected to go beyond your first tingling/itching/burning warning signal. “Remember...Research is the key to YOUR cure!” Each Step Toward Finding an Effective Treatment is Important! Contact the APF on 1-866-APF-3635 or email porphyus@porphyriafoundation.org.

### RECORDATI RARE DISEASES AND PANHEMATIN®



Recordati has updated the dosing and administration video to reflect the new 350mg vial. You or your healthcare professional can view this step-by-step tutorial on how to administer PANHEMATIN® to patients, including dose calculations here: <https://www.panhematin.com/healthcare-professionals/dosingimage.png>

### ALNYLAM RECEIVES FIRST EVER FDA APPROVAL TO TREAT RARE DISEASE hATTR AMYLOIDOSIS



Alnylam Pharmaceuticals received FDA approval for the first ever RNAi therapeutic, Patisiran, that treats patients with a rare disease known as hereditary ATTR amyloidosis. RNAi is a biological messenger that takes the coding from DNA and carries those genetic instructions around the body - silencing or interfering that message can halt or slow the body's production of the problematic protein found in hATTR patients. As a result, patients can finally get the relief they need. The FDA approval was made possible due to positive phase 3 results which met both on the primary and secondary endpoints. The study showed that patients treated with Patisiran displayed significant and clinically meaningful improvements in measures of polyneuropathy and quality of life. Alnylam Pharmaceuticals is also hard at work completing the phase 2 and phase 3 clinical trials for Givosiran for the treatment of acute hepatic porphyria. Many of our APF members are currently participating in the Phase II and Phase III clinical trials for Givosiran. Patients can finally get the relief they need.

### GIVOSIRAN PHASE THREE CLINICAL TRIAL RECRUITMENT COMPLETE

Givosiran is an investigational RNAi therapeutic for the treatment of acute hepatic porphyrias (AHPs). Thank you to every patient that is able to participate! You are a medical hero! We all appreciate your dedication to helping everyone with AHP.

### SCENESSE NEW DRUG APPLICATION FDA FILING COMPLETE!

Clinuvel Pharmaceuticals Ltd. has filed a historic New Drug Application with the FDA for the use of SCENESSE® in EPP. The submission includes data from five clinical trials in EPP, Compassionate Use, Special Access Schemes, and real-world experience from European use – that is nearly 6,700 afamelanotide doses administered to more than 800 patients over 12 years. Clinuvel has requested Priority Review to the FDA, which is the quickest possible review process. The final review will begin once the data has been validated by the FDA, which will result in a target date for



CLINUVEL

completion of the review in either 6 months (priority review) or 10 months (traditional review). The APF has met with the FDA on multiple occasions, initiated the Scientific Workshop held in October 2016, and continues to advocate for the approval of SCENESSE®!

#### US TIMELINE

2005 Clinuvel initiated communication with FDA	2018 June 22 – New Drug Application submission by Clinuvel
2008 Orphan Drug designation granted by FDA	2018 August 22 – New Drug Application validation target date
2010-2013 Phase 2 and 3 Clinical Trials	2019 Approval OR Complete response by FDA
2016 Fast Track designation awarded by FDA	

## THE ROLE OF OUR RESEARCH COORDINATORS

If you have had an appointment at one of the eight porphyria centers around the country – or any facility that participates in research – you have been introduced to the role of a research coordinator. The research coordinator acts as the glue between the patient, the physician, and the sponsor of a research study or clinical trial. They organize, manage, and control the many aspects of the patient interaction in a study. Each of the six porphyria centers and two satellite centers has research coordinators organizing the patient experience in a study or trial. Take a moment to understand their role, thank them, and help them do their job well by being compliant in a study! They are critical to the research process, and help the patient community turn research into potential treatment and cures!

## MEET DEE FAUST, RESEARCH COORDINATOR AT WAKE FOREST UNIVERSITY



Dee Faust has worked at Wake Forest University for over 20 years – the last three with Dr. Herbert Bonkovsky, Porphyria Expert. She was enamored with the role of interacting with patients and collecting data. Her desire to work with patients in this medical area became a reality when a former supervisor recognized her ability and hired her as a data collector – and that's all she wrote! A typical day for Dee includes recruiting patients, organizing study visits, receiving labs, and ensuring everything is timely and that nothing falls through the cracks. She contacts pharmaceutical companies with questions, reaches out to patients, works on new studies, prepares budgets, enters data... everything A-Z! Communicating with physicians is a very important part of her job, making sure to keep them fully abreast of what is going on with each study and each patient. Dee mostly enjoys the study visits, and being the calm in the storm. Patients often come to a center feeling frustrated, scared, and angry. Porphyria has affected their life in every shape and form. To her, that initial phone call is probably one of her favorite moments - "They've hit a brick wall again and again – no one has listened to them talk, actually heard them and understood their challenges." She thrives on shifting angry, upset and tearful meetings into positive information gathering sessions. Dee thinks "what if it was me, my mama, or my child." Budgets are not her favorite, but it's sure part of getting the studies done. "Today's research is tomorrow's medicine" is her mantra. Dee would like patients to know that their role is the most important – being compliant, letting her know information about medications, procedures, appointments. It's a new world for a lot of people, so a simple call asking "how's your week going" is important. In fact, every patient has her cell number – so they have a way to connect at all times. She tells every patient, "don't just have it, use it!" When she isn't taking care of patient needs, she enjoys time with her husband and daughters, the beach, and camping (in an RV, not a tent!), and watching movies. Dee wants all to know that she loves the APF. "I love everything the APF stands for – I mean that from the bottom of my heart." We love you too, Dee!

## CALL TO ACTION

Thank you to all who answered the call to write letters to the FDA requesting priority review. They received 511 letters from APF members and patient advocates describing the burden of living with EPP and requesting PRIORITY REVIEW for SCENESSE®. The ENORMOUS stack of advocacy letters landed on the desks of decision-makers, once again showing the unmet medical need for a treatment for EPP. Still want to write a letter? Send your plea to approve Scenesse to:



Scott Gottlieb, M.D., FDA  
Commissioner, Office of the  
Commissioner, U.S. Food and Drug  
Administration, 10903 New Hampshire Avenue, Silver  
Spring, MD 20993.

## TAKE ACTION ON FACEBOOK



Head to the APF groups on Facebook to show your support for the approval of Scenesse. You can join this closed group by request at APF: Porphyria- EPP. Can't wait to see you there! #ApproveScenesse #APF #EPP #EPPLIFE



REMEMBER...RESEARCH IS THE KEY TO YOUR CURE!!!

## DRUG QUESTIONNAIRE FOR ACUTE PORPHYRIAS (AIP, VP, HCP, ADP)

The APF recently sent out a mailing to our patient members requesting to share information about the drugs that you are currently taking. The American Porphyria Foundation is collaborating with researchers to identify new safe and unsafe drugs. We need YOUR help. Are you experiencing adverse effects with any of your new medications? Did you receive this questionnaire? If so, please fill it out and return it to the American Porphyria Foundation. If you did not receive this questionnaire, please email Edrin Williams, Director of Patient Services to request a copy at [edrinw@porphyriafoundation.org](mailto:edrinw@porphyriafoundation.org). We will share these results with our team of renowned Porphyria experts/researchers. They are in the process of updating our safe and unsafe drug list for the acute porphyrias. Your donations will help us educate physicians about the dangerous effects of unsafe drugs.

## IMPORTANT SURVEY FOR ACUTE AND PCT PATIENTS

The American Porphyria Foundation has recently developed surveys for both the acute hepatic porphyrias (AIP,VP,HCP or ADP) and PCT. We are seeking better ways to understand what YOU are enduring! We are trying to overcome some of the stigma associated with porphyria, such as being called a drug seeker or hypochondriac. This survey will increase our awareness on what our patients experience once diagnosed and how physicians manage their care. These surveys are expected to arrive in your mailboxes mid-October. If you agree to participate, please return this survey by mail to the American Porphyria Foundation. If you did not receive this survey, please contact Edrin Williams, Director of Patient Services by phone at 301.347.7166 or by email at [edrinw@porphyriafoundation.org](mailto:edrinw@porphyriafoundation.org) to request a copy to be sent to you.

## CAMP SUNDOWN 2018 – PATIENT EDUCATION MEETING

Camp Sundown 2018 was a special experience for all who attended. After 23 years, this was the first year that an entire week of camp was devoted to EPP. Families hailed from both the US and Canada (Thank you Canadian Association for Porphyria for sponsoring two families to attend!). Days and nights were flipped and every moment was filled with fun and friendship for both the children and parents. Life is magical when the sun is not a concern! The APF held a patient education meeting during the second full day of camp – parents gathered together while the children ran around without a worry about sun exposure. It was wonderful week and a valuable experience for all! Thank you, Camp Sundown!



**“I cannot express in words the experience and the value for our young EPPers and their families to spend a week where they don’t have to worry about the sun. This is an amazing place! Lifelong friendships are made here for both the parents and the kids. Thank you to all that make it possible for my young EPP Brothers and Sisters to live one week of a “normal” life.”**  
**#PorphyriaWarriors**

**~ Rob Saupe on Camp Sundown**



## EPP AND CAMP SUNDOWN FEATURED ON FOXNY



EPP and Camp Sundown were highlighted in a segment on Fox News NY on Thursday, August 2nd, titled The Big Idea: Camp Sundown. You can find the segment if you search “Fox NY and Camp Sundown” – you may recognize some faces! APF member and EPP patient Morgan McKillop spoke eloquently about living with EPP and the joy of going to camp where she is safe and has a wonderful time at all times of day and night! View at <http://www.fox5ny.com/news/350839189-video>.

## PATIENT EDUCATION AND SUPPORT MEETINGS

Hosting a Patient Education and Support Meeting in your local community is a valuable opportunity to bring patients, caregivers and supporters together to share common issues and to learn more about porphyria from an expert. Discussions include medical explanations, information about current and emerging treatments, and an extended time for Q&A to answer specific questions with the help of a porphyria expert. Bringing together a group facing similar issues in a social atmosphere is a meaningful and educational way to help individuals understand that they are not alone. We are here to help you coordinate your efforts. If you are interested in hosting a Patient Education and Support Meeting, please contact Edrin Williams, Director of Patient Services at the APF. Meetings are being scheduled in the following locations with volunteers who have graciously offered to host.

**UPCOMING MEETING:** Cincinnati, OH hosted by Audrey Schering and Brandy Nicole Smith – Saturday, September 29, 2018. | San Francisco, CA - November 2018 | San Diego, CA - December 2018

**TENTATIVE LOCATIONS:** Cromwell CT, Tampa, FL, Philadelphia PA, Seattle WA, Myrtle Beach SC, San Francisco CA, San Diego CA, Loma Linda CA, San Antonio TX, Oklahoma City OK, Birmingham AL, Bethlehem PA, Milwaukee WI, Boston MA, and Cook Station MO.



## PLAY BALL!!

An amazing group of basketball enthusiasts held a tournament to benefit the APF in early August. Nick Guanciale, APF member, organized the 1st Annual Marc Guanciale 2 on 2 street basketball tournament to benefit the American Porphyria Foundation. The idea blossomed with the idea to have a few friends over to play, but quickly grew into a mission to back the organization that supports his rare disease. At the event, Nick spoke about the importance of the APF to him, including the focus on children through the new Shadow Jumpers and Light the Moment programs. Nick was diagnosed with EPP when he was 13. Thank you, Nick!!

## PORPHYRIA PET CALENDAR! 2019



Stay tuned for a fun way to raise awareness for porphyria while putting your favorite pet in the limelight! The APF will be launching a 2019 PET CALENDAR event. The calendar, which will be ready for sale in mid-November, will include porphyria facts and details – alongside photos of your favorite family pets. It is often our loving and reliable pets that help us through the hard times as we manage through the pain, isolation, and even loneliness that can be part of living with porphyria. This calendar will celebrate these animals in our lives. Start getting your shots of your cute canine, fabulous feline, or even your gorgeous ghecko ready to submit! No professional photos, please. Any submitted photo should only include your pet – no humans. All types of pets will be eligible, and each submitted pet will be included in the calendar – though several will be selected to be featured through a fun contest. We hope this will be a creative way to share the joy of our animals give to us AND create porphyria awareness. Stay tuned for the program to be launched in September!

**HAVE AN IDEA FOR A FUNDRAISER?** Call the APF.  
We assist in events supporting porphyria!

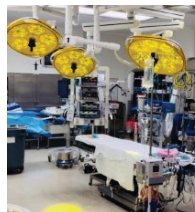
## LIVER TRANSPLANTATION Q&A

### Why do people with Porphyria have Liver Transplants?

Liver transplants are performed to treat liver disease in EPP and to treat the intractable neurologic symptoms of the acute porphyrias. It is also performed in PCT patients with liver disease secondary to hepatitis C and alcohol abuse. AIP patient, Karen Eubanks, (LT and kidney transplants) and young EPP patients, Madelyn (LT) and Skylar (LT) have all had a liver transplant. Although rare, in some cases, EPP patients develop excessive production of protoporphyrin in the bone marrow which damages the liver and causes end-stage liver disease. Also, patients can have liver damage secondary to protoporphyrin. At that point they need a life-saving LT. However, the genetic defect in EPP is expressed in erythroid cells, where LT will not correct enzyme deficiency.

Liver transplant is not an easy undertaking. In both EPP and acute porphyria, pre- and post-transplant patients can develop neurologic dysfunction, proximal motor weakness, respiratory paralysis, hypertension, tachycardia, pain, nausea, vomiting, biliary complications, hepatic artery thrombosis, and a host of severe symptoms, including prolonged ventilation. During the transplant, photosensitive porphyria patients can suffer skin and tissue burns. Protoporphyrin is activated by light in the 400 nm range. Special filters are used to block light below the 470nm and provide protection from phototoxic injury while still allowing the surgeon a good surgical view. Also difficult, the transplant is often followed by a regimen of immunosuppressant medications, steroids and other transplant drugs. Unfortunately, the genetic defect in the bone marrow is not corrected by LT, so liver disease can recur. Thus, there have been cases of re-transplantation. In the acute porphyrias, some patients are not responsive to treatment or no longer have venous access. In these instances, LT is used to quell these frequent, life threatening attacks. In the acute porphyrias, LT can reverse the enzyme defect and correct the genetic defect in AIP, thus restoring PBG and ALA levels. These are the neurotoxins that are thought to affect the nerve damage. The enzyme defect is not corrected in EPP during LT. Acute and EPP patients should be referred to a Porphyria Center of Excellence to help with the pre and posttransplant care. Liver transplant is a very serious surgery and requires multiple tests, psychological evaluation and careful consideration.

## AORTIC VALVE REPLACEMENT AND CORONARY ARTERY BYPASS GRAFTING IN A PATIENT WITH EPP



APF member and bypass recipient, Robert Doyle, shared his story with the APF about his physicians that went above and beyond to protect him from harmful lighting during his procedure. Robert developed heart issues unrelated to his EPP, which required bypass surgery. His physicians – Lu Zheng, MD and Donna Kucharski, MD, Department of Anesthesiology, Allegheny Health Network, Pittsburgh, PA – developed his case experience into a poster that was presented at Allegheny Health Network. Intraoperatively, they utilized yellow light filters ordered

from an industrial light supplier. The surgeon noted no significant impairment to his vision. Maintenance of optimal hemoglobin level through blood prime of the CPB bypass circuit was used to avoid over activation of heme synthesis. The surgery was performed smoothly and the patient was discharged in good health without any acute episode of phototoxicity. WE OFFER OUR GRATITUDE TO PHYSICIANS THAT GO THE EXTRA MILE FOR OUR PATIENTS!



## INTERNATIONAL CONGRESS ON PORPHYRINS AND PORPHYRIAS (ICPP) PATIENT DAY 2019

The dates have been announced for the next ICPP to be held in Milan, Italy. The Congress will take place at the University of Milan from September 8-11, 2019. "Patient Day" will be held on Sunday, September 8th, offering the opportunity for patient-physician interaction and patients' empowerment. An opening lecture on Sunday evening will open the scientific program.

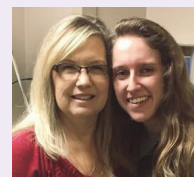
## CANADIANS ACCESS PANHEMATIN®



The Canadian Association for Porphyria is very pleased to share that their advocacy to have the Canadian Blood Services (CBS) distribute the treatment for those with acute porphyria has been successful. Mel Cappe, CBS Chair, is supportive of CBS carrying Panhematin® and is committed to expediting the process. Although there is still a process within CBS to be followed, they are committed to making Panhematin® available through their system. This will make it possible for Canadians with acute porphyria to finally have equitable access to their treatments as required. There were three presentations to the CBS Board that helped to move them toward providing access. These are available on the Canadian Association for Porphyria website: <http://canadianassociationforporphyria.ca>. We offer our congratulations to our neighbors to the north – access to Panhematin® is critical.

The American Porphyria Foundation is proud of your hard advocacy work to attain this necessary access to life-saving treatment.

## KAREN EUBANKS – LIVER TRANSPLANT UPDATE



APF member Karen Eubanks recently underwent a liver and kidney transplant. Karen was diagnosed with

AIP in 1999, enduring all the symptoms. In 2009 she began receiving Panhematin weekly. However, she began to lose kidney functions due to her porphyria, going into Stage 5 renal failure. On June 19-20, Karen received a liver and kidney transplant at the University of Arkansas for Medical Sciences in Little Rock, AR. It was a 12-hour surgery which kept her in the hospital for six days, with her remaining recovery at home. After 19 years she was told that she is Porphyria Free! Karen says, "It's a big decision but I was at peace with it because I put it in God's hands. I couldn't fathom getting treatments and dialysis every week to survive."

## PAIN IN THE ACUTE PORPHYRIAS BY DR. KARL ANDERSON, UTMB



Pain is the major symptom of an acute attack of porphyria. The pain is severe, especially in patients sick enough to seek treatment at an emergency room or hospital. The pain almost always requires treatment with a narcotic analgesic. Characteristically, the pain is: diffuse in the abdomen rather than localized steady and unremitting; accompanied often by pain in the back, chest, extremities and elsewhere, and constipation, or less commonly diarrhea not associated with peritoneal signs such as tenderness, guarding or rebound; accompanied often by signs of ileus such as distension and decreased bowel sounds.

In a patient with well-documented acute porphyria, the diagnosis of an acute attack is clinical, and not a laboratory diagnosis. Other causes of abdominal pain should be considered, especially if the symptoms are different from previous episodes. However, pain and

other manifestations, such as fluid and electrolyte imbalances, can be treated while other causes are being excluded. Adequate doses of a narcotic analgesic should be given at intervals frequent enough to control the pain.

There is virtually no risk of addiction, since acute attacks usually respond well to treatment with Panhematin®. Therefore, severe pain and the need for narcotic analgesics are short-lived. Some patients with frequent attacks of porphyria have chronic pain between their attacks. When they present with an acute attacks, such patients may already be taking a narcotic analgesic for chronic pain, and their regimen may be guided by a specialist in treating chronic pain. Increased tolerance to narcotics may have developed and higher doses may be required for acute pain control. After recovery from an acute attack, such patients are expected to return to their usual pain regimen. It is rare for patients with acute symptoms of porphyria to have true drug-seeking behavior.

## FDA MEETING ON PATIENT FOCUSED DRUG DEVELOPMENT FOR CHRONIC PAIN



In July, the FDA hosted a meeting on on Patient-Focused Drug Development for Chronic Pain. The FDA is interested in hearing patients' perspectives on chronic pain, views on treatment approaches, and challenges or barriers to accessing treatments for chronic pain. FDA is particularly interested in hearing from patients who experience chronic pain that is managed with analgesic medications such as opioids, acetaminophen, nonsteroidal anti-inflammatory drugs (NSAIDs), antidepressants; other medications; and non-pharmacologic interventions or therapies. You can

access the meeting webcast here: <https://collaboration.fda.gov/pfdd070918>. Take the opportunity to comment on the Public Docket: <https://www.regulations.gov/document?D=FDA-2018-N-1621-0001>.

## APF PAIN PROGRAM

Porphyria patients repeatedly are placed in demeaning positions about the need for pain medications at their Primary Care Doctors Clinic, Emergency Rooms, and Hospitalists. Too often, physicians have sadly withheld appropriate pain medication and treatment so stop the acute attack. If they experienced the intensity of the pain, we believe they would administer treatment immediately. Below are descriptions from patients during an acute attack:

- I felt as if a thousand flaming swords were stuck in my abdomen and left there to burn without relief.*
- If you want to see a grown man cry, no, scream in pain, see me in an attack.*
- It is like a hot, steeltoed boot kicking in your stomach and never stopping.*
- My whole abdominal area is on fire; burning, raging hot fire!!!!*

The pain of acute porphyria can last for days, weeks, months. Generally, because of the safety issue of certain pain medications, narcotics are given to relieve pain. Since the use of narcotics establishes a host of medical issues and questions, the suffering patient is often left suffering until the need for a narcotic is firmly settled. Such an attitude extends the pain unnecessarily and is demeaning to the patients.

Here is what these same patients have been told about their pain.

- When the doctor left the room, the nurse said, "You can't fool me. I know all about you drug wanting people." I cried the rest of the day. My husband could not console me.*
- I have never been a person who wanted to take drugs or drink. Therefore, accusations of being a drug addict infuriates and saddens me, because the pain is so devastating. They are not true but there is nothing I can do about it. It is a hopeless feeling.*
- I take my APF ER kit to the ER and hospital with me every time. Some doctors are very interested and take time to look at it and others won't even take it out of my hand. The result is that I either get treated or I don't, depending on who sees the information.*

While many doctors choose to "watch and wait" until the attack subsides before administering treatment, patients suffer in excessive pain. Stopping an attack, should help stop the symptoms, including the extreme pain. By allowing the attack to continue to see if glucose works is almost inhumane to patients who are suffering miserably. The APF would like to enlighten doctors as to the severity of porphyria pain and assist patients to receive treatment sooner.

## ACUTE PORPHYRIA PATIENTS NEED OPIOIDS

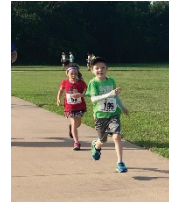
Although we understand that the misuse of and addiction to opioids has become a national crisis affecting public health, as well as social and economic welfare, we need to protect citizens who live in intractable pain. Many of these people are unable to function without the respite that opioids provide. The Acute Porphyrias are a group of ultra-rare metabolic diseases which are known among the most painful in humankind. Furthermore, the acute porphyrias are pharmacogenetic diseases, meaning a genetic inability to properly metabolize many drugs exists, thus causing dangerous, life-threatening reactions. Opioids are the only group of pain management drugs that can be prescribed safely to treat porphyria pain without placing patients' lives in severe danger. Therefore, it is imperative that porphyria patients, who are compliant with their prescribed instructions, be given a special exclusion to receive opioid prescriptions with the assessment of their own physicians who understand the porphyrias. Our Scientific Advisory Board of porphyria experts who are renowned worldwide in the field, and all of are keenly aware of this impending problem that patients will endure undue suffering without understanding.

## PORPHYRIA J AND THE SPARTAN RACE



A self-described family man and health enthusiast, Jared also lives with EPP. You may know Jared from his YouTube videos, PorphyriaJ, available on the APF YouTube channel, where he creates awareness about EPP. Jared ran 3+ miles and overcame 23 daunting obstacles in the Boise Spartan Sprint on behalf of Porphyrians everywhere. The Spartan race motivates people to get out of their comfort zones and grow through resiliency. In order for Jared to complete this race - he tested every ounce of his resilience. For Jared, this race is an outward expression of a deep internal struggle that has developed through a lifelong challenge of avoiding sun exposure. He wants all who suffer from this disease to know they are not alone and to believe that positivity is created by taking care of yourself and focusing on what you can do. Jared says 'Seek Shade and Stay Happy' - a motto he lives by and shares with all who suffer along with him. Jared raced in support of the AMERICAN PORPHYRIA FOUNDATION...thank you!

### HERE'S WHAT JARED INSPIRED ...



**FB Post July 4:** "Clint pushed himself today and ran a 1 mile race in the sun and heat. So proud of him!! Thank you Jared Ulmer. We watched your video Monday and you really inspired him!! #EPP (We would usually be more covered but we stayed in the shade except for the race. He is feeling good and not showing signs of pain!)"



**FB Post August 9:** "My husband has EPP and is terrified of doing outdoor activities with the kids). A few weeks back he found a YouTube blog by a guy named Porphyria J. He was inspired by a video of this guy with EPP doing a spartan race like a BOSS! There was a link at the bottom of the video listing all the protective clothing available on Amazon so I bought it ALL for him. Here is a picture of him with our kids on holiday!"



### NEW PUBLICATION: PATIENT PERSPECTIVE ON AIP

*Patient Perspective on Acute Intermittent Porphyria with Frequent Attacks: A Disease with Intermittent and Chronic Manifestations* was recently published in *The Patient*, authored in part by Desiree Lyon Howe and Jessica Hungate. Although clinical aspects of acute intermittent porphyria attacks have been documented, the experience of patients is not well known, particularly for those more severely affected patients who experience frequent attacks. The aim of the present study was to qualitatively characterize the experience of patients with acute intermittent porphyria, as well as the impact of the disease on daily living. In this study population of acute intermittent porphyria with frequent attacks, most patients had symptoms during and between attacks. In these patients, acute intermittent porphyria appears to have acute exacerbations as well as chronic day-to-day manifestations, and is not just intermittent as its name implies. As a result, patients reported limitations in their ability to function across multiple areas of their lives on a regular basis and not just during acute attacks. For the full article, visit <https://link.springer.com/article/10.1007/s40271-018-0319-3>.

### PHYSICIAN EDUCATION TRAINING IN ACTION!

A major initiative of the APF is physician education. APF member and Variegated Porphyria patient, Sharon Dill, recently experienced our outreach first-hand. An amazing thing that happened during a recent hospital stay...

"One of the many healthcare providers involved in this hospital stay, is a Pedro Llera, MD Hospitalist. When he introduced himself on Wednesday, he told me he was very interested in AHP (Acute Hepatic Porphyria) and wanted to know what I could tell him. He truly wanted to learn from me, as I was his first ever AHP patient. He told me that this year he has started a new diagnostic process. If a patient has recurrent, unresolved and undiagnosed abdominal pain, nausea, vomiting, diarrhea, constipation, he has them tested for AHP! He has

not had someone with it yet, but believes it is the right thing to consider.

Today, I finally got a chance to ask Dr. Llera how he got so interested in AHP. He said in medical school, those few minutes on it really intrigued him and his best friend, Dr. Jose Vasquez. His best friend now works at another hospital in town. They frequently discuss AHP. He told me his friend recently attended a conference on AHP and brought back all sorts of great information - and that's when they both started to include in potential diagnosis. As we talked I learned it was the physician education conference held this past January in Orlando, FL. This is the same meeting that I attended and presented at on Patient Day!

I know the APF works so hard to put on and support these educational opportunities - and they are working! Two more docs are thinking Porphyria! And because of this knowledge, Dr. Llera has been able to be a great advocate for me. Thank you, thank you, thank you!!! Sharon



### CONGRATULATIONS TO APF STAFF MEMBER NATALIA!

Natalia Sturza has worked for the APF for several years and continues with special projects related to computer work. She has wonderful news to share! Natalia was recently granted US citizenship. She has been an asset to the APF and has worked hard in school and other positions - and now has a new role as a citizen of the US. Please join us in congratulating Natalia on her accomplishment!!



# SHADOW JUMPERS UPDATE *Light the Moment* 2018

## *Success at Disney for the Stuhlsatz Family*

In March, we selected our first LIGHT THE MOMENT recipient, The Stuhlsatz family! "Light the Moment" is a Shadow Jumpers Program that helps EPP families go on vacations usually viewed as "too sunny." For our first year, we sent Clint and Hattie along with their parents to Walt Disney World all-expenses-paid. With extended park hours into the late night, protective clothing and fast passes during the day, as well as thought into where to stay in the resort, Disney can be done right, just a little different. We also got a private VIP to help minimize time walking around the park! Huge thanks to some sponsors for donating; Delta Airlines, Solombra by Sun Precautions, Blue Lizard and Therma Freeze. The entire trip was possible due to the generosity of wonderful donors!

**Hattie (age 4):** "The trip was so fun. My favorite ride was the Slinky Dog ride. I really liked meeting the princesses."

**Clint (age 6):** "I had so much fun on the rides and wanted to go again. My favorite ride was a spaceship ride that you got to it in and control the buttons and gears. They had really yummy food too!"

**Allison and Ben (kids at heart):** "It exceeded our expectations. It felt like you were in a whole different world. The trip planning was excellent from start to finish, with minimal effort needed on our part. It was a once in a lifetime opportunity. Kirk, our VIP guide, was one in a million. He went above and beyond getting us specialized treatment, and assuring we saw and did any activity that the kids wanted. Even more important, he was very conscientious of the kids' EPP. He kept them in the shade and had UV protective umbrellas to keep them blocked when walking about. He was so considerate, fun, and knowledgeable. He made the experience something we will never forget. We ate like royalty while there and got to experience so many activities that we never thought would be feasible for our family. Initially the kids would feel a little self-conscious like normal but as the fun began, they got so sidetracked by the excitement and thrills of being at Disney, they quickly forgot about their cover-ups and enjoyed being a kid!"

## RARE ON THE ROAD – HOUSTON



APF Director of Patient Services Edrin Williams attended Rare on the Road – A Rare Disease Leadership Tour hosted by the Every Life Foundation and Global Genes on Saturday, June 9. Rare disease patients, advocates and caregivers gathered together to gain new skills and insights to take advocacy efforts to the next level. This interactive and engaging experience gave advocates a chance to learn how to navigate your community through active engagement. The tour for this year has concluded. Stay tuned for next year's events. They may be coming to a city near you!



## YOU MADE IT THROUGH SUMMER!

You did it. You survived the hottest, stickiest months of 2018. It wasn't easy! You geared up, you hid, you shadow hopped, and you avoided that big round ball of fire in the sky. Sometimes, you suffered – both physically and emotionally. It wasn't easy, but you are an inspiration! Here are some brilliant ways APFers – both cutaneous and acute members - survived the summer sun...



Bailey's garage setup!



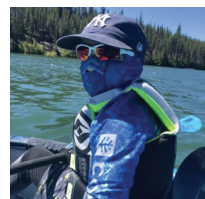
Creative shoes



Shawn Willis



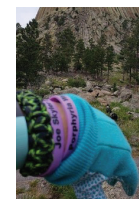
Jessica Rutlege



On the water



Pool shade



Hiking



Rob Saupe



**DONATIONS TO APF**  
[porphyriafoundation.org](http://porphyriafoundation.org)  
 or 1-866-APF-3635

## IN MEMORY & IN HONOR

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

### IN MEMORY:

Dr. Karl Anderson, Megan Parish and Jennifer Ewing for **Richard J. Howe, PhD.**; Ariel Ikesakes for **Daniel Pudlicki**; Carol Kuklewski for **Vince Kuklewski**; Maragert Haggerty for **Carol Rusnak**.

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

### IN HONOR:

Kathleen Angela Shiel for **Ralph Gray**; Carole and Ken Aitchison for **Cameron Wheeden**; Sharon Koch for **Jagger and Jake Liguori**; Linda Nagin for **Melisa Nagin**; Leigh Brooks for **Vivian Martinez**.

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.

All information and content on this Web site are protected by copyright. All rights are reserved. Users are prohibited from modifying, copying, distributing, transmitting, displaying, publishing, selling, licensing, creating derivative works, or using any information available on or through the site for commercial or public purposes.

What's New at the APF  
[www.porphyrifoundation.com](http://www.porphyrifoundation.com)



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

Address Service Requested

4915 St. Elmo Avenue, Suite 105, Bethesda, MD 20814

AMERICAN PORPHYRIA FOUNDATION



Non-Profit  
U.S. Postage  
PAID  
Houston, Texas  
Permit No. 7279