



AMERICAN PORPHYRIA FOUNDATION

1st Quarter, 2019



April 6-13, 2019

PORPHYRIA AWARENESS WEEK - APRIL 6-13, 2019

The challenge of living with porphyria starts with how little is known about it among friends, family and the medical community. That is why **PORPHYRIA AWARENESS WEEK** is very important. **PAW** provides each of you with the opportunity to enhance porphyria awareness in your local and medical communities. We hope that you work to engage in your local communities to educate about Porphyria. We would love for each of you to be involved in this week in some way – and will help you create a successful effort, big or small! The APF has developed a toolkit,

available on the APF website, that will supply you with ideas and items for the week. The APF office can supply you with brochures, porphyria fact sheets, information to gain media attention and so much more. We want YOU to get involved this year!

STAY TUNED FOR UPDATES, TOOLS, AND PROGRAMS SPONSORED BY THE APF! #APFPaintTheWorldPurple #PAW2019

NEED IDEAS FOR YOUR COMMUNITY?

FOR ADULTS:

- Set up an exhibit table at a local hospital, doctors' office, community center.
- Create traveling displays or interactive exhibits and present at local community centers, libraries, and other public spaces.
- Engage your local media representatives.
- Host a fundraiser for the American Porphyria Foundation.
- Run a series of interviews on your website, social media or in your newsletter with researchers from your organization or institution. Consider videoing portions of the interviews and posting them on your website or YouTube channel.

FOR CHILDREN:

- Organize a day when an entire class wears purple for porphyria.
- Set up a PAW display in your school with material about porphyria.
- Organize a Walk for Porphyria at your school.

"NON-EVENT" PAW CELEBRATIONS:

- Use social media! Share APF posts or start your own.
- Share Porphyria Awareness Week information with your local, state, and/or national government official(s).
- Write an article or editorial about porphyria for a local publication.
- Blog or write about PAW on your company website.
- Write letters to the editors of your local newspapers.

Share your Porphyria Awareness Week effort with the APF. Send pictures and stories to Edrin Williams, Director of Patient Services, at edrinw@porphyriafoundation.org.

PROTECT THE FUTURE TRAINING

Karl Anderson, MD, UTMB, Galveston, TX, led a *Protect the Future* physician training in February. Six physicians from across the country attended the mini-course and were trained under the world-renowned Porphyria expert. The APF is proud to support this training and will now have additional resources for our patient community. The training is critical to ensure that the expertise of our experts is transferred to young physicians. We were thrilled to support training for Amy Dickey, MD (Massachusetts General Hospital), Natalia Dixon, MD (Wake Forest), Annie Prah, FNP (Wake Forest), Mohammed Kazamel, MD (University of Alabama Birmingham), Alice Cusick, MD (University of Michigan) and Ting Li, MD (Wake Forest). We are grateful for the contributions to the *Protect the Future* program that enable this training.

APF WEBSITE – BOOKMARK AS A "FAVORITE"



Please take a moment to bookmark the **APF** website as one of your favorites! You will have easy access to valuable content including the Safe/Unsafe Drug List

for Acute Porphyrias, events, recent news, clinical trials, APF Merchandise, Member Stories and so much more! Already have the site bookmarked? With the launch of the new website this Fall, you will need to re-bookmark the site www.porphyriafoundation.org.

FDA GRANTS PRIORITY REVIEW TO SCENESSE®



CLINUVEL

On January 10, Clinuvel Pharmaceuticals Inc. announced that the US Food and Drug Administration Division of Dermatology and Dental Products set a Prescription Drug User Fee Act (PDUFA) date of July 8, 2019 for SCENESSE®. The drug was granted Priority Review, which indicates an unmet medical need for treatment. A New Drug Application for the use of SCENESSE® (afamelanotide 16mg) in adult patients with erythropoietic protoporphyria (EPP) had been finalized June 22 with the request for Priority Review. SCENESSE® is a controlled release injectable implant containing the active ingredient afamelanotide. CLINUVEL conducted five clinical trials of SCENESSE® in EPP. Two randomized, placebo-controlled clinical trials of SCENESSE® conducted at US EPP expert centers showed the drug enabled patients to increase the time spent outside without experiencing phototoxicity and improved quality of life.

What is a PDUFA date? Originally passed in 1992, PDUFA authorizes the FDA to collect fees from drug manufacturers to fund the drug approval process. Review deadlines begin on the date that a new drug application is accepted by the FDA.

What is Priority Review? Under PDUFA, the FDA is usually given 10 months to review a new drug application. However, if a drug is designated for priority review, the FDA is given six months to review that drug.

YOU DID IT!!! Your hard work writing letters, making calls, attending meetings and making your voice heard at the FDA had a significant impact on the determination to grant Priority Review to Scenesse. Thank you to all the wonderful Porphyrin advocates that advocated for this treatment!



SAFE / UNSAFE DRUG DATABASE SURVEY

The American Porphyrin Foundation mailed surveys to our members with acute porphyria (AIP, VP, HCP and ADP) regarding Safe/Unsafe Drugs and the adverse effects that it had on them. The purpose of this survey was to gather information on drugs that may have affected you. We took this information to our team of renowned Porphyrin experts/researchers to identify new safe and unsafe drugs. This survey will also be used to evaluate our current Safe/Unsafe Drug Database. We appreciate all the submissions that we received. If you did not receive a Safe/Unsafe Drug Questionnaire, please contact Edrin Williams, Director of Patient Services at the APF Office (866-APF-3635) to request a copy.

GIVOSIRAN - PHASE 1 CLINICAL TRIAL RESULTS



Alynlyam Pharmaceuticals announced that results from the Phase 1 study of givosiran, for the treatment of acute hepatic porphyria (AHP), were published in The New England Journal of Medicine (NEJM). Monthly givosiran administration resulted in sustained reductions of ALAS1 mRNA, urinary ALA, and PBG to near normal levels. These reductions were associated with a 79 percent decrease in mean annualized attack rate, compared with placebo. Topline results from the complete 6-month double-blind portion of ENVISION Phase 3 trial are expected in early 2019. Rolling submission for a new drug application (NDA) has been recently initiated, with full clinical sections planned to be submitted in mid-2019, assuming positive results. <https://www.nejm.org/doi/full/10.1056/NEJMoa1807838>



PANHEMATIN® RECENTLY LAUNCHED IN CANADA

The Canadian Association for Porphyrin has been hard at work advocating for the approval of PANHEMATIN® for Canadian citizens living with acute porphyria. Recordati Rare Diseases Canada Inc., a biopharmaceutical company providing orphan therapies for patients with rare diseases, recently announced the commercial launch of PANHEMATIN®, the first prescription medication approved by Health Canada for the treatment of recurrent attacks of acute intermittent porphyria (AIP). We are hopeful that our Canadian friends will have similar access to this life-altering treatment that the US has held since it was approved in the US nearly forty years ago. Though Health Canada has granted approval for PANHEMATIN®, patients continue to struggle with access. It is currently available to order through hospitals, though the effort for broadscale coverage continue. It is estimated that there are between 1 and 2 patients per 100,000 with the disease in Canada, which would translate into 360-720 Canadians with AIP. In 2016 and 2018, the Canadian Association for Porphyrin surveyed Canadian porphyria patients to evaluate their access to hemin treatment. The results for those diagnosed with an acute porphyria and told by a physician that they should receive hemin showed that only half of those had access to treatment. "We are excited that PANHEMATIN® is now available for Canadian patients as a treatment for painful and devastating AIP attacks," said Renata Sklodowska, M.D., Director Medical Affairs, Recordati Rare Diseases Inc. "The fact that a disease is uncommon does not mean that it should go untreated. Recordati remains committed to supporting patients with rare disorders, including the needs of the Canadian AIP community." The Canadian Association for Porphyrin will continue to advocate until all Canadian patients have timely access to PANHEMATIN®!

CLINUVEL NEWS FOR VARIEGATE PORPHYRIA (VP)

In October 2018, CLINUVEL announced that it has reached agreement with two European porphyria expert centers on a clinical trial protocol to conduct a Phase II proof of concept study evaluating the safety and efficacy of SCENESSE® (afamelanotide 16mg) in variegate porphyria (VP). The study (CUV040) will start patient treatment in the northern hemisphere Spring of 2019. VP is classified as both a cutaneous and acute porphyria. The cutaneous symptoms are characterized by blistering and chronic fragility of sun and light-exposed skin, especially the back of the hands and the face.

INTER-AGENCY TASK FORCE ON PAIN and THE OPIOID CRISIS

The Pain Management Best Practices Inter-Agency Task Force is an advisory group overseen by the U.S. Department of Health and Human Services and the departments of Veterans Affairs and Defense. Its 29 members include federal officials and pain experts from academia and private practice, as well as a patient advocate representative from the U.S. Pain Foundation. This task force was convened to help manage the effect of the opioid crisis in the US. Their recommendations will provide guidance for pain management for physicians across the US.

Why does this matter to Porphyria? The acute porphyrias are pharmacogenetic diseases – drugs that are unsafe trigger potentially life-threatening attacks. The ONLY safe drugs that manage the pain of an acute attack are opioids. Our community needs to have a voice in ensuring that the task force understands the severe pain of acute porphyria and demands that patient-centered care is a benchmark in these guidelines. Patient-centered care ensures that each patient is treated individually and not lumped into the ineffective pain management guidelines for other diseases.

ABOUT THE REPORT The task force has released a much-anticipated draft report with recommendations for improving the management of chronic and acute pain. The public is being asked to provide feedback on the report by April 1. Then the report will be finalized and submitted to Congress. Key takeaways from the report:

1. Pain is an **enormous public health problem** with profound individual and societal consequences.
2. Successful management of **pain requires individualization of care** in the selection of therapies tried, in the consideration of risks and benefits of therapies, in the duration of treatment, in the optimal dosing of medication and so on.
3. Best practice in pain management is achieved through a **multi-modal, multidisciplinary, integrated model of care** with a range of pharmacological and non-pharmacological treatments.
4. **Stigma is a major barrier to treatment**, so it is critical to provide education and awareness of the underlying process of pain and provide empathy and a nonjudgmental approach to treatment.
5. **Public, patient, and provider education is critical** to the delivery of effective, patient-centered pain management and is necessary for optimizing patient outcomes.
6. The risk-benefit balance for opioid management **must be considered on an individual basis** as there is wide variation in factors that affect the optimal dose of opioids.

NOW IS THE TIME TO SPEAK UP!

Here's what to do by April 1:

1. **Write a letter and tell us your pain story. include your diagnosis, describe your pain, say that you cannot use unsafe drugs, address your letter to:**

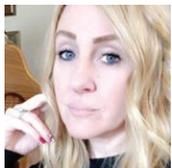
U.S. Department of Health and Human Services
Office of the Assistant Secretary for Health
200 Independence Avenue, S.W., Room 736E,
Attn: Alicia Richmond Scott, Task Force Designated
Federal Officer
Washington, DC 20201

2. **Send your letter to the APF office via mail or email to EDRINW@PORPHYRIAFOUNDATION.ORG. we will collect all letters and mail them together. Together we are strong!**

American Porphyria Foundation
4915 St. Elmo Avenue, Suite 105
Bethesda, MD 20814

WE HEAR YOU!

APF member Tara Cantley wrote the following during a recent attack. She said what many with a chronic disease feel. Can you relate?



Thank you for your courage, Tara.

"Let me make something clear.

1. *I can't just get up and "feel better."*
2. *I'm chronically ill, meaning it will not go away.*

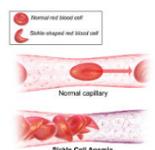
3. *I can't just "find something to do" and forget about it.*

4. *I can't help the way I feel and what this illness does to my body.*

5. *Let me rest as long as it takes to recover.*

You can't expect someone to be hospitalized and be 100% two days later. It doesn't work that way. I WILL feel better eventually but I WILL also go through this again many times, but I HAVE. NO. CONTROL. Stop making me feel bad over something that I can't stop/control."

SICKLE CELL DISEASE and ACUTE PORPHYRIA



The APF was asked by the government Pain Task Force to participate in a conference call with the Sickle Cell Organization to discuss our mutual problem of extreme pain. Sickle cell disease is an inherited blood disorder marked by flawed hemoglobin. Sickle cell, like the acute porphyrias, is an excruciating group of diseases that involve heme. Normally, hemoglobin allows red blood cells to carry oxygen from your lungs to all parts of your body. Normal red blood cells are smooth, rounded, flexible and can flow freely. In Sickle Cell disease, when these red blood cells lose their oxygen, they become rigid, sticky and misshapen, like a sickle, and live only 10 days as opposed to the normal 120 days. These sickle-shaped cells obstruct capillaries and restrict blood flow to an organ resulting in pain and other serious damage. The crises are extremely painful and often require pain management with opioids. Sharing our mutual problems to pain management was helpful to the pain task force. Please remember to send your own personal story about your pain and difficulty gaining appropriate pain management.

INTERNATIONAL CONGRESS ON PORPHYRINS AND PORPHYRIA 2019



Every two years, an international congress is held to bring together the scientific leaders in the porphyria community, and to host an international patient day. This year, the meeting will be held in Milan, Italy with the theme *From Bench to Care*. It will be a wonderful place to discuss exciting topics on the Porphyrins and learn about recent progress. Patient Day will be held on Sunday, September 8th, organized by the

patient's associations, offering the opportunity for patient/physician interaction and patient empowerment. This is a wonderful opportunity for patients and advocates from around the world to meet and learn about recent advances in Porphyria. The opening lecture on Sunday evening will begin the scientific program followed by a welcome reception at the magnificent cloister of the University of Milan. The official program will start Monday with basic science on fundamentals on Heme, Iron and Heme regulation, moving to acute and chronic Porphyrins including novel information on pathophysiology and treatment. Oral and poster presentations will be throughout the program featuring the work of young physicians and scientists. A major focus of the conference will be "how to define an acute attack of Porphyria" with the goal of collaborating to reach a consensus during the conference. The program will ensure critical attendance of young physicians and scientists at this conference. To learn more about the ICPP 2019, please visit www.icpp2019.org.

WHY ENZYME TESTING IS NO LONGER USED FOR BASIC DIAGNOSIS OF ACUTE PORPHYRIAS

A number of patients have asked why experts are no longer testing enzyme activity for acute porphyrias. According to experts, DNA is now the "gold standard" to discover a porphyria mutation. Enzyme measurements are not as sensitive or specific as DNA analysis and are rarely used for diagnosis for any type of porphyria.

DNA defines the mutation (your type of porphyria) but does not define if you are in an attack. Instead, for initial screening while you are ill, a spot urine sample is obtained for measurement of urine PBG, ALA and total porphyrins. If none of these are elevated, acute porphyrias can be excluded as a cause of recent or concurrent symptoms. PBG measurement is most important and specific for acute porphyrias. However, PBG and ALA may be less elevated and return to normal more quickly after an attack of Variegate Porphyria or Hereditary Coproporphyrinemia than in Acute Intermittent Porphyria but do elevate. Therefore, measurement of total urine porphyrins is important, keeping in mind that a very high elevation of urine porphyrins can occur in many other medical conditions and are not used alone to diagnose.

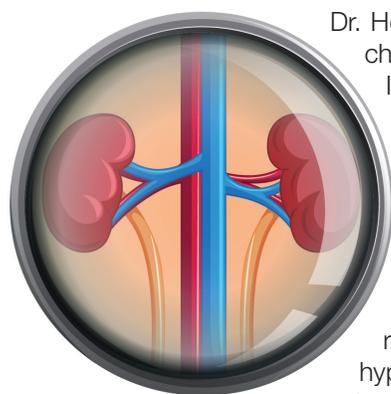
The measurement of specific heme-synthesis enzyme activities is considered a second-line test in evaluating porphyrias. Enzyme measurements are often used for identifying a genetic trait for porphyria in the family of a diagnosed person. Enzyme activity measurements are also used when a symptomatic person is diagnosed with porphyria of some type, but the pattern of heme-precursors does not allow differentiation of the type of porphyria. Measurements of enzyme activity otherwise have limited utility in the evaluation of suspected porphyria in symptomatic individuals.

CPOX/Hereditary Coproporphyrinemia (HCP), is a particularly labile enzyme after removal from the body. Vigorous specimen processing or failure to properly maintain the specimen during transport or storage can cause enzyme damage and produce falsely low (i.e. false positive) results. The measurement of specific heme-synthesis enzyme activities is commonly considered a second-line test in the evaluation of porphyrias. Enzyme activity measurements are most often used for identifying a genetic trait for porphyria in family members of a person diagnosed with porphyria. Enzyme activity measurements can also be useful when a symptomatic person is diagnosed as having a porphyria of some type, but the pattern of heme-precursors does not allow differentiation of the specific type of porphyria.

An assay for CPOX in whole blood was offered in the 1990s but resulted in many false-positive results and ultimately was discontinued. Reductions in enzyme activity do not necessarily reflect a genetic trait; certain exogenous agents, such as alcohol or lead can inhibit or interfere with the activity or alter the synthesis of specific enzymes. There is typically a substantial degree of overlap in enzyme activity values for normal individuals and for individuals with a porphyria characterized by deficiency in that enzyme. Therefore, measurements of enzyme activity otherwise have limited utility in the evaluation of suspected porphyria in symptomatic people.

Measurements of urinary PBG/ALA, fecal and plasma porphyrins are essential for diagnosis of clinically overt porphyrias because symptoms cannot be ascribed to porphyria unless specific patterns of overproduction of porphyrin precursors/porphyrins are demonstrated. Enzyme measurements may mislead due to overlap between normal and disease ranges. DNA, urine, fecal and plasma porphyrins are part of testing.

KIDNEY DISEASE AND ACUTE PORPHYRIA



Dr. Herve Puy and his colleagues at the French Porphyria Center enhanced knowledge of the biological characteristics of renal pathology in their studies of the cellular mechanisms of Chronic Kidney Disease. In 2003, 415 AIP patients in the study were followed by the French Porphyria Center researchers. A follow-up was conducted in 2013, assessing patients for clinical, biological, and histological (the branch of biology which studies the tissues of animals and plants) parameters. Chronic kidney disease occurred in up to 59% of the AIP patients and was a long-term complication, which can lead to end stage renal disease (ESRD). Development of ESRD decreases urinary excretion of PBG and ALA, further elevates plasma levels, and may increase symptoms. Plasma porphyrins may increase sufficiently to cause blistering phototoxicity/photosensitivity. Combined liver and kidney transplantation can correct the metabolic symptoms of this hepatic porphyria and restore renal function. Since individuals with chronic AIP can develop long term complications, such as hypertension, kidney damage potentially resulting in kidney failure, and a form of liver cancer known as hepatocellular carcinoma, it is suggested that liver and kidneys be included on your regular check-ups.



HAVE YOU READ THE PURPLE LIGHT BLOG?

<http://blog.porphyrifoundation.org>

NEW MEMBER STORIES

The APF website features the stories of members who have shared their personal experience with Porphyria. The feedback we receive is astounding from current and new members alike – reading the stories of others help them feel less isolated and better understood. We hear “it’s not just me!” almost every day. Sharing a personal experience also has an impact on the storyteller. It is a form of journaling your experience, one that is poorly understood by many. We invite you to share your story. You don’t have to be an expert writer – we will share in the editing process and can help you write a story that you are comfortable sharing. Contact Edrin Williams at edrinw@porphyriafoundation.org to start the process.

M. McNutt, PCT “I consider myself to be a pretty confident, healthy, and strong woman with a whole lot of life to live. Life is beautiful, and I never knew how much I took it for granted until ...” *Read more at <https://porphyriafoundation.org/member-stories/murphy-mcnutt>*

J. Reeves, EPP “I have experienced symptoms of EPP for most of my life without knowing what was causing them. My symptoms gradually got more severe as I...” *Read more at <https://porphyriafoundation.org/member-stories/john-w-reeves-iii>*

A. Butt, CEP “When I was born, I was completely normal like other children. When I was 2-months-old, my mother cut my nails and there was starting water from it. When the nails’ water touches my face, it...” *Read more at <https://porphyriafoundation.org/member-stories/abdul-waheed-butt>*

THE LORD IS MY SHEPHARD



Terri Witter, APF member (AIP), has a wonderful collection of sheep that has blossomed over the last forty years since her diagnosis. Terri was given her first stuffed lamb along with a card that read “This is to remind you of who you are and who is taking care of you.” Now she has nearly 250 sheep ranging from soap and butter to ceramics and cake pans! Sheep have given her courage on her socks while speaking at the FDA, comfort while hanging on her IV pole in the hospital, and a good giggle when she added iron-on sheep to her undies when headed for a hospital visit! “The Lord is my Shephard, I shall not want. Each goofy little lamb is my reminder that I am in God’s care at all times,” shared Terri.

FINDING THE UNEXPECTED



Many years ago, Dr. Selig Epstein, a porphyria researcher, published a high impact paper on acute porphyria titled “The Prevalence of Acute Intermittent Porphyria” (George D. Ludwig and I. Selig Epstein). Normally, that would be the story in the APF newsletter. However, this research took an unpredictable turn. The lab technician participating in the study demonstrated how to test for acute porphyria. The unexpected happened...the test was positive! The lab tech had Acute Intermittent Porphyria and did not know!



PANHEMATIN® PREVENTION STUDY

The APF is actively seeking patients who receive prophylactic heme treatment that has been effective in preventing acute attacks. Are you currently on a regimen of PANHEMATIN® infusion? Call the APF to learn more. It is critical that we find patients to participate in this study. Your travel/hotel will be managed for you, and you will be under the expertise of Dr. Karl Anderson, UTMB, Galveston, TX.



PUBLICATIONS ALERT

BENEFITS OF PROPHYLACTIC HEME THERAPY IN SEVERE ACUTE INTERMITTENT PORPHYRIA

To read the full report visit: <https://www.sciencedirect.com/science/article/pii/S2214426918301289>

Dr. Bonkovsky, Porphyria Expert, Wake Forest University, led a recently published case study on the use of prophylactic heme therapy in severe Acute Intermittent Porphyria. Recurrent attacks of AIP, >3 attacks in a year, with at least one attack requiring intravenous heme therapy, are associated with significant morbidity, lost productivity, and health care burden. The report finds that prophylactic heme therapy can potentially decrease the incidence of acute attacks and associated issues including quality of life and healthcare costs.

ACUTE HEPATIC PORPHYRIAS: REVIEW AND RECENT PROGRESS

To read the full article: <https://aasldpubs.onlinelibrary.wiley.com/doi/full/10.1002/hep4.1297>

This recent publication from our Porphyrias Consortium has just appeared in the on-line journal of the AASLD: Hepatology Communications. The article provides an update about diagnosis and management of the acute hepatic porphyrias.

TREATMENT OF DISABLING PORPHYRIA

HemOnc today We are grateful to Dr. Samuel Silver, University of Michigan, who recently published an article regarding the difficulties in diagnosis, management and treatment of the acute porphyrias. The article discusses the disabling aspects of living with Porphyria including misdiagnosis and access to effective treatment. To read the full article: <https://www.healio.com/hematology-oncology/hematology/news>

EPP FEATURED IN STAT NEWS

An article featuring APF member, Jennifer Beck, was recently published in STAT News. The article highlights Jennifer’s struggle with Erythropoietic Protoporphyrinemia and her quest for treatment with Scenesse. The article touches on the impact of living with EPP. Clinuvel, the APF, and patients living with EPP agree that, as Desiree Lyon said, “it’s well past time for this drug to win FDA approval.” You can access the full article here: <https://www.statnews.com/2019/01/02/new-treatment-may-help-people-who-are-allergic-to-the-sun/>

ATTENTION, SHADOW JUMPERS! Who wants to go to Disney?



FIND YOUR SHADOW, 2019

Send in your application now for the opportunity to be selected to win an all-expenses paid trip to Disney! You will find all information and the application on the APF website at www.porphyrifoundation.org/patients/shadow-jumpers/find-your-shadow. The Find Your Shadow program was created to help families impacted by EPP go on vacations they may think are "too sunny" to attempt. Vacations with EPP can be difficult to think about and hard to plan. We wanted to make this a bit easier for one family every year! The Shadow Jumpers team will work to plan all aspects of this vacation...and send a child and their family on the trip of a lifetime! This program is funded through private donations.



SHADOW JUMPERS COMIC BOOK

Your Shadow Jumper will love this comic book, written by a porphyria expert and a genetic counselor. Designed for Youth ages 5 and up or for the adult young at heart. This book has captivating content and vivid display colors. Both fun and informative, the storyline features children with EPP on mission to solve crime. Written By- Shrutti Shenbagam - Hetanshi Naik, PhD - Manisha Balwani, MD, Illustrated by- Tina Chu.



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. AHP - How do you balance a healthy diet where you don't gain weight OR can lose weight and still maintain sufficient carbs to keep a porphyria attack at bay?

A. The utility of carbohydrates in treatment of acute attacks is still unclear. Carbohydrates may be helpful at the start of an acute porphyria attack, when symptoms are still mild, in stopping the attack from getting more severe. The evidence for this effect is still inconclusive because quality clinical trials to test this have not been done. On the other hand, there is little to no evidence to show that taking carbohydrates has been shown to prevent an acute porphyria attack from starting. So, I do not recommend taking carbohydrates on a regular basis to prevent acute attacks.

Q. How frequent do you have someone come across with a dual porphyria? Why is this important to know? Many say they have AIP/VP, AIP/PCT or AIP/EPP.

Each of the genetic porphyrias are rare, and it is exceedingly rare to find people who have disease-causing mutations that lead to symptoms in two different porphyria related genes, for example AIP and EPP. It may be more likely for someone to have PCT in addition to one of the other porphyrias, particularly an acute hepatic porphyria. PCT is the most common of the porphyrias and most often are caused by inhibition of the UROD enzyme within the liver. Because it is much more common, symptomatic PCT has been reported in patients with another genetic porphyria.

Can we please post an answer on iron supplements for EPP?

A. There are two porphyrias that present clinically with severe, acute photosensitive, EPP, caused by mutations in the ferrochelatase (FECH) gene and XLP caused by mutations in the ALAS2 gene. In patients with EPP due to FECH mutations, too little iron may further decrease the function of ferrochelatase, potentially making the symptoms worse. The idea is that in EPP patients who also have too little iron, iron supplements may help improve symptoms. This is currently being studied by the Porphyrins Consortium.

Why, in CEP, is there nothing for patients but complete isolation from sunlight and certain lights? Why is there not an SPF or medications we can use? What is suggested?

A. While all cutaneous porphyria patients should practice sun avoidance, it is critical for CEP patients to avoid sunlight because the damage due to sun-exposure is much more severe in CEP patients. Unlike other porphyria patients with blistering skin lesions (such as PCT), CEP patients are much more likely to have their damaged skin get infected, ultimately leading to irreversible skin damage. In addition, CEP patients are at risk for permanent eye damage that can lead to blindness and permanent teeth discoloration with sunlight exposure. Unlike in EPP, where patients develop acute symptoms that serve also as warning signs of too much sun exposure, CEP patients have minimal acute symptoms that warn them of too much exposure. While reflecting sunscreens that protect the skin from all forms of light are partially effective in protecting the skin of CEP patients, they do not offer complete protection, and for CEP patients, this can give a false sense of protection and lead to too much sun exposure and irreversible skin and eye damage. For these reasons, we recommend complete avoidance of sunlight for severe CEP patients.

RARE DISEASE WEEK

RARE DISEASE WEEK

ON CAPITOL HILL

Edrin Williams, Director of Patient Services, attended Rare Disease Week on Capitol Hill. This week consisted of a Legislative Conference, Lobby Day and a Rare Disease Congressional Caucus briefing. The Legislative Conference brings patient advocates together to learn about legislative issues and about building relationships with members of Congress. Patient advocates visited their representative offices to fight for research funding, Medicare/Medicaid, access to treatment, bills to be passed and, more importantly, to share their powerful stories. As a constituent, you have the right to make sure that your voice is heard. Stay tuned for information regarding In-District Lobby Days. Let's Lobby for Porphyria!

RARE DISEASE DAY – FEB 28

The American Porphyria Foundation had the opportunity to exhibit at the National Institute of Health on Rare Disease Day. This conference was widely attended by patients, patient advocates, rare disease organization leaders, health care providers, researchers, industry representatives, and government employees. Director Kristen Wheeden was present to educate everyone about the mission of the APF, Patient and Physician Education programs, awareness efforts and legislative and federal initiatives.

LIVING THROUGH HER ART



Sandra "Pinky" McKinney-Humphrey, 67, recently died after a long illness and a lifelong battle with acute porphyria. During her life, her hands and creative spirit were her gift and she used them well into retirement. She viewed art as a natural process that began with her eyes, emanated through her heart and to her hands. From writing poetry to wood carving, ceramics, sculpturing, drawing, and painting, she amassed a vast collection of representations of people, places and abstract forms. Her family displayed her art at her funeral as a fundraiser for the APF. Friends and family donated to the APF as they selected art from her collection. We are grateful for the blessings received from Sandra.



PORPHYRIA STRONG: RESPONDING TO PORPHYRIA WITH RESEARCH AND EDUCATION IN BOSTON, MA



In January, Amy Dickey, Elin Storjord, and Padmini Pillai presented at gastroenterology (GI) grand rounds at Massachusetts General Hospital (MGH), a teaching hospital of Harvard Medical School. For each of them, porphyria is personal. Amy is a critical care physician at MGH who has EPP. Elin is a physician in Norway who is getting her PhD on the topic of AIP. While she previously had AIP, she was cured by a liver transplant several years ago. Padmini is a PhD in immunobiology who is completing her postdoctoral research in immunoengineering at Massachusetts Institute of Technology. She has AIP. At GI grand rounds, they shared personal experiences, taught about porphyria, and discussed recent research, including Elin's own research findings. After presenting, they spent the week brainstorming research ideas, sharing experiences, and showing Elin around Boston. Drawing on their personal experiences and scientific training, this team of researchers is putting their heads together to understand porphyria and to find treatments. They are hoping to work together to find new answers for porphyria. Stay tuned to see what is next for this Boston strong trio! (Photo l-r: Amy Dickey, MD, Padmini Pillai, PhD, Elin Storjord, MD)

IN MEMORY & IN HONOR



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

IN MEMORY:

Byran Staley for **Karen Nicewarner**; Constance McLean Reese for **Sandra McKinney-Humphrey**; Valerie Vashio for **Sandra McKinney-Humphrey**; Kelly Thompson for **K. Wiggins**; CarolAnn and Paul Stickler for **Dick Howe**; Cantigny Research Found. /Diane Levere for **Dr. Richard M. Levere**; Donna Payton for **Dusty Parker**; Zila Reichman for **Tomy Reichman**; Cerkoney, Stauff, Oleary, Stewart for **Fred Cerkoney**; Kathleen Giacobbe for **John H. Giacobbe**; Stephanie Rushwin for **Suzette Frazzini**; Desiree Lyon for **Luca**; Anita Nagler for **Astairre Dever**; Christine Baer for **Richard M. Chiles**; Gary Horn for **Susan Horn**; John and Barbra Stark for **Astairre Dever**; Dale Brehm for **George Brehm**; Davida Hansen for **Annamae McCarthy**; Kathleen Toelkes for **Donna Pagnano**; William Lambert Jr., William Lambert III, Carol Smith, Eve Soltau for **Norma Kelton Lambert**; Lani Longshore for **Karen Nicewarner**; Theresa Avella for **Karen Nicewarner**; Judith Federline for **Karen Nicewarner**; Eric Brown for **Karen Nicewarner**; Michael and Carol Farina for **Vincent K. Farina**; Susan Kenney for **Vivian Martinez**; Dianna Poissant for **Matthew Cole**; Keith and Eletheer Decker for **Karen Nicewarner**.

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

IN HONOR:

Sophie Foucault for **Fredrick Michael Cerkoney**; James and Barbara Witter for **Terri Witter**; Christin Steiskal for **Tracy Nudo**; James Peetz for **Desiree Lyon**; Rachel Wise for **Jere and Pauline Wise**; Kathleen Venter for **Patricia Wright**; Carol Gaudette for **Tristen Gaudette**; Jennifer Ewing for the **APF**; Elaine Sasso for **Elizabeth Britton**; Thomas Jacobs for **Candace Johnson**; Mary Frances Donnelly for **Ralph Gray**; Martin and Connie Helleson for **Jennifer Streeter**; Lois Aitken for **Griffin Vasquez**; Cheryl Harriman for **Jeffery Pradovic**; Dale Moczynski for **Alicia Moczynski**; Ginger Steinhilber for **Fern McLean and Kristen Steinhilber McGlaughlin**; Kathleen Toelkes for **Linda Peterson and Valine Jensen**; Myrna Cartledge for **Ralph Gray**; William Gray for **Ralph Gray**; Maureen Curran for **Desiree Lyon**; and Greater Cincinnati Foundation for **Patricia Wright**.

PATIENT EDUCATION AND SUPPORT MEETINGS

The APF will host a dozen Patient Education and Support Meeting across the US in 2019. These 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. Locations: March 23, Ann Arbor, MI | April 4, Seattle, WA | April 27, Placentia, CA | May 31, Salt Lake City, UT | Birmingham, AL | New York City, NY | Winston Salem, NC | Chicago, IL | Boston, MA | Orlando, FL.

To host 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**.

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What's New at the APF
www.porphyriafoundation.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.

Pet Calendars 2019



The APF would like to thank all participants who submitted a photo of your adorable pets for our Pet Calendar Contest! The contest was a huge success, allowing the APF to help train the next generation of porphyria experts through the *Protect the Future* program. Every photo that was submitted was featured in the calendar. It's not too late to order! APF Pet Calendar 2019 are available for purchase by contacting the APF office through autumnlee@porphyriafoundation.org or through the APF store by following the link <https://porphyriafoundation.org/store>. A special thank you to APF member Nicole Castellano for her hard work in helping the APF create this calendar!

April 6-13, 2019

PORPHYRIA AWARENESS



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