AMERICAN PORPHYRIA FOUNDATION

SUMMER 2025 **NEWSLETTER**

IN THIS ISSUE

PORPHYRIA AWARENESS WEEK

This year, Porphyria Awareness Week (PAW) aligned with Global Porphyria Day for a united, worldwide effort to raise awareness. Together with our global partners in the Global Porphyria Advocacy Coalition (GPAC), we joined forces to shine a brighter light on porphyria. PAW 2025 ran from May 12 to May 18, culminating in Global Porphyria Day on May 18. Through out the week, the American Porphyria Foundation (APF) hosted a variety of activities—but the most impactful efforts were those organized by our members, our porphyria community. Patients know firsthand what it means to live with this condition, so their voices were key to raising awareness and driving change. This year's theme was THINK PORPHYRIA . We helped our medical teams remember to THINK PORPHYRIA when they are confronted with porphyria symptoms. *Carlos Lopez pictured above created a fantastic Awareness Event at the Woodland Park Playground in San Diego County. The event was supported by Truly Nolen Pest Control. Carlos has been hosting a series of awareness events in honor of his family who suffer with AIP Many thanks, Carlos.* The PAW agenda was an exciting one that reached the medical community and the public:

HAT DAY: Families, friends, patients, caretakers wore hats in support of those with photosensitive forms of porphyria, such as EPP, XLP, CEP, PCT, VP and HCP. Schools participated by hosting HAT DAY and asked students to give presentations to their classmates.

WEAR PURPLE DAY: A simple yet effective way to spark conversations about porphyria was wearing pur-ple, especially with the APF logo. Some women even painted their nails purple.

PORPHYRIA PET DAY: Pets wore their APF bandanas or other purple wear.

SHARE YOUR STORY DAY: Patients and Caretakers posted their experiences, which is a timeless way of engendering hope and friendships.

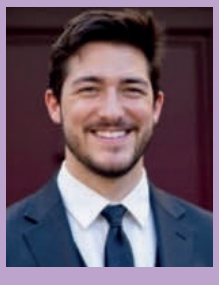
LIGHT THE SKY PURPLE: Spearheaded by Kelly Burns, a Canadian VP patient, this campaign illuminated iconic landmarks across Canada and the U.S in purple light to honor porphyria patients. Hopefully, you can get the lights turned purple for next year to light up the world.

APF PODCAST: Host Andrew McManamon and guest, Mike Boone, shared his amazing acute porphyria journey as a research patient and one on both Panhematin and Givlaari to quell the horrific attacks . He shared his most recent cancer battle and the way he keeps porphyria at bay.

GLOBAL PORPHYRIA DAY and **SATISFY THE SOUL SUNDAY** Friends around the world shared their lives and hopes while enjoying Satisfy the Soul Sunday.

THINK Porphyria

APF Podcast



Our popular, ***Rarely Discussed***, Podcast is viewed around the world. Producer, Andrew McManamon, hosted Mike Boone and Kelly Burns in the most recent Podcasts. To View ***Rarely Discussed***, Mike and Kelly, as well as the many other podcasts with friends from around the world visit: *https://beacons.ai/rarelydiscussed*. Guests include patients, doctors, experts, caretakers, and the other parties interested in porphyria who have an amazing story.

If you would like to share your experience as a porphyria patient, caretaker of member of a medical team, please contact the APF at 866-APF-3635.

Mike Boone A Medical Hero

In late 2002, after a yearlong battle, I was diagnosed with Acute Intermittent Porphyria (AIP). At first, I was relieved to finally have answers. I wasn't the first in my family to be diagnosed. In the late 1950's, my great uncle was diagnosed and died of the disorder. My grandmother would follow in the early 60's and died at the young age of 28. My mother was also diagnosed in the mid 1990's.

Needless to say, I thought I understood the seriousness of the situation, but after learning what my treatment options were and experiencing so many attacks, I became nervous. How could I support my family. I quickly found the American Porphyria Foundation/APF. Through them, I learned about Panhematin and jumped at the chance to have my first infusion. After years of horrendous pain and illness and repeated attacks of AIP, I finally had my life returned to me. I wanted to give back, so I began joining research studies. The first study I enrolled in was the Longitudinal Study, which was straight forward. It required no travel and only one time did I have to send any samples. It was a simple blood draw at my local doctor's office and then mailed to Salt Lake City. I have remained in the study and once in a great while, I fill out more pa-pers with questions about how I am doing.

The second study I took part in was the Panhematin study in Galveston, Texas at the University of Texas Medical. This study required me to fly to Galveston, Texas and stay in the hospital for four days. As with most trials, the research grant paid for all of my travel expenses and all study related hospital expenses. The APF arranged all the travel. Again, this study had a simple procedure. Once I began feeling as though an attack was coming on, I contacted the APF and they made the travel arrangements. The next day I flew into Houston where a car service picked me up and drove me to UTMB. This study was a double-blind study, which means that neither I, nor the doctors and nurses knew if I was getting the Panhematin or a placebo. Only the pharmacist knew. I had to wear a blindfold, the tubing and medicine was wrapped in foil, and a sheet was also placed between me and the IV and bottle. This happened once a day for four days, and I was also on normal saline with 10% dextrose constantly while I was there. Things went smoothly and before I knew it, I was on my way back home to Colorado feeling good that I had helped my fellow patients.

The next trial was for Givosarin, a then new treatment for acute porphyria that reduces the number of attacks. Unfortunately, I got the placebo instead of the drug, but I persevered because having placebo results for a research trial is as important as the real drug. Now I am on Givosarin/Givlaari, which has reduced my attacks and when I have breakthrough attacks, I have Panhematin infusions to stop them in their tracks. Between the two drugs, I was able to become employed again and return to a normal life.

Recently, my life has been interrupted with a cancer diagnosis. To make matters worse, one of the treatments caused me to have a porphyria attack. Again, I contacted friends at the APF and they not only provided a concerned shoulder to lean on, but they also helped with the attack. I am in treatment waging a new battle with confidence. I encourage all of you to join a research trial for the benefit of all.

Editor's note: Our prayers are with You Mike. Thank you for your years of service as a research volunteer. You are our Medical HERO.

KELLY BURNS

Kelly began experiencing symptoms early in life—persistent nausea, nerve pain, abdominal pain, light sensitivity, and an extreme sensitivity to medications. Her journey to a diagnosis took an astounding 31 years. Because her symptoms were vague and often misdiagnosed, Kelly's father dismissed them as hypochondria, while her mother suspected something was wrong but believed it was driven by anxiety. Unfortunately, this experience is all too common for those living with acute porphyria. As time passed, Kelly's condition wors-ened. She endured the excruciating abdominal pain that is a hallmark of acute porphyria, and she also suf-fered from severe photosensitivity, which complicated her diagnosis even further.

At age 39—31 years after her symptoms began—Kelly finally found a doctor who was willing to look beyond the usual suspects. They ran screening tests for PBG and ALA, which revealed a significant accumulation of both porphyrin precursors. The doctor called with the long-awaited news: Kelly had been diagnosed with acute porphyria. Further testing confirmed the specific type—Variegate Porphyria (VP), which impacts both the central nervous system and the skin's sensitivity to light.

To manage her attacks, Kelly now takes Panhematin and is hopeful about the future availability of Scenesse in Canada to help treat her photosensitivity after the VP research is complete. But Kelly hasn't just focused on her own recovery—she has become a powerful patient advocate. She joined the board of the Canadian Association for Porphyria (CAP) and launched what many believe is the most impactful porphyria awareness campaign to date: the *Light the Sky Purple project*.

Continued on next page.

MEMBER STORIES

Thanks to Kelly's hard work, this inspiring initiative has illuminated over 80 public and private landmarks, bridges, fountains, skyscrapers, stores, homes, and more—with purple lights in recognition of Rare Disease Day and Global Porphyria Day. Purple is used as porphyria is named after the Greek word *porphyrus* — meaning purple as reddish-purple urine often occurs in acute attacks. We invite everyone to join this movement. Ask your local businesses, city councils, friends, and neighbors to light the sky purple. Our dream is that one day the world will be bathed in purple light in solidarity with porphyria patients everywhere.

ADVOCACY NEWS

Exciting News For EPP and XLP Friends - Bitopertin Research Trials

disc)medicine

The new Bitopertin Clinical Trials Are Now Enrolling for EPP and XLP--Clinical trials are now open to evaluate the safety and effectiveness of bitopertin in treating erythropoietic protoporphyria (EPP) and X-linked proto-porphyria (XLP) in individuals aged 12 and older. Bitopertin, the Disc Medicine treatment also known as Disc-1450, will be researched at various study sites across the country with more sites opening in time. The study aims to answer two key questions:

- **Does bitopertin increase pain-free sunlight exposure after six months of treatment?**
- **Do PPIX concentration levels change from before bitopertin treatment to after 6 months of treat-ment.**

Participants will receive either bitopertin or a placebo (a look-alike substance with no active drug). They'll complete daily questionnaires and attend regular study visits for monitoring and assessments. Travel ex-penses and trial related expenses will be paid by the sponsor. For details including the eligibility criteria, please contact the APF. If you are interested in joining the trials, please contact the APF at 866-APF-3635 or *pax@porphyriafoundation.org*. Joining the trial is very simple, and the result can be life changing !! To read about the trials, see: *https://www.clinicaltrials.gov/study/NCT06910358?cond=EPP&term=bitopertin&rank=1*

ATTENTION HEALTH CARE PROFESSIONALS Disc Medicine will be presenting a new educational plat-form about EPP and XLP for Health Care Professionals. Dr Bruce Wang and Dr. Gayle Ross provide an overview of EPP/XLP in 3-part on demand educational program. To learn more, see: *www.medlive.com/v/EPP*

Health Canada Validates Scenesse Submission

Our Canadian friends will be interested that SCENESSE has been submitted as a new drug. Thanks Canadian EPP patients, Ian McCool and Kelly Burns, and to the Canadian Association for Porphyria for their efforts to help Canadian patients gain access to SCENESSE and to educate them on present trials for Variegate Porphyria.

Recordati Rare Disease Click Campaign

Thank you Recordati makers of Panhematin! The APF received a $5000 donation from Recordati for their PAW click campaign. Every click on the Home Page of the APF Website engendered a $5.00 donation to the APF. The Recordati logo includes Focuses on the Few because rare disease is the core of their work. Recordati works side-by-side with rare disease communities to raise awareness, educate, and advocate for improved diagnosis. The click campaign was one such effort. Thank you friends at Recordati Rare Disease!!!

Mitsubishi Tanabe Pharma America

Results from a first-in-human study of dersimelagon, an investigational drug for EPP. A Study to Investigate the Safety, Tolerability and Pharmacokinetics of MT-7117 in Healthy Subjects, NCT02834442

Results from a first-in-human study of dersimelagon, an investigational oral selective MC1R agonist - Pub-Med

https://pubmed.ncbi.nlm.nih.gov/37060458/

DNA Testing Program

As part of the Alnylam Act service for patients and physicians, they provide tree DNA testing for acute porphyrias from Prevention Genetics. The APF includes these forms in our comprehensive Doctor Packet. It is essential to note that to gain access to the test, patients must give the form to their doctors or provide the doctor's name and contact information for the APF to send the form directly to the physician. The doctors must fill out the form, sign and submit it by fax to Prevention Genetics. The eligibility criteria includes, fami-ly history, elevated urinary PBG and ALA, unexplained recurrent (more than one), prolonged (>24 hours) epi-sodes of severe, diffused, poorly localized abdominal pain and at least two of the following symptoms, Red to brownish urine, Blistering skin lesions on sun-exposed areas, Peripheral nervous system manifestations occurring around the time of abdominal pain (i.e., motor neuropathy [paresis], sensory neuropathy [numb-ness, tingling, limb pain, Central nervous system manifestations occurring around the time of abdominal pain (i.e., confusion, anxiety, seizures, hallucinations), Autonomic nervous system manifestations occurring around the time of abdominal pain (i.e., hyponatremia [Na in blood < lower limit of normal], tachycardia, hypertension, nausea and vomiting, constipation). Please also note that DNA testing identifies the mutation but does not denote if a person is in an attack. Thus, biochemical testing/screening is essential for acute porphyrias. For further information, contact the APF at 866-APF-3635.

ATTENTION **Please NOTE:**

The complimentary DNA forms must be signed by a physician and submitted to Prevention Genetics. Unfortunately, some individuals have contacted Prevention Genetics directly and have been unable to access the test.

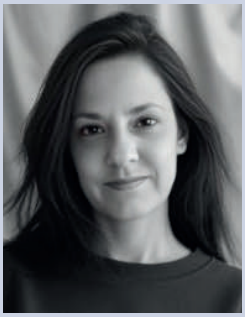
Please ensure that the forms are provided to your physician for their signature and submission.

If you would like a packet with free DNA forms, please contact the APF toll free at 1-866-APF-3635

ANNOUNCEMENTS

Want To Know About APF Social Media Groups? Contact AMY BURKE

If you have questions about or would like to join our any of our eight FACEBOOK groups, Instagram, and Twitter , contact our social media coordinator , Amy Burke on FACEBOOK or AmyB@porphyriafoundation.org. Amy joined the APF when she faced a Porphyria Cutanea Tarda (PCT) diagnosis.



During the summer of 2009, Amy began noticing dark urine and pain. Her doctor said, "I don't know what you are eating or drinking to make your urine this color," so Amy decided to try to drink more water and see the results. A few weeks later, she started to develop large water blisters on her fingers and hands, as well as a rash on her forearms. Her doctor's salve treatment did not work, so she tried a dermatologist who thought Amy had PCT and took a biopsy of her lesions. The test was shipped to Mayo for testing and after a week of waiting, the diagnosis was confirmed as familial PCT. Her blood work also showed that Amy had suffered Hepatitis B at one time and her liver enzymes were also extremely high.

The dermatologist explained PCT and scheduled a phlebotomy and careful monitoring of the liver function. Her office tried to contact a hematologist to get things started; it took over 3 weeks before a doctor finally took the case. Her doctor admitted that he did not know a lot about the disease. Her first treatment was terrible! Her blood pressure dropped to 43/70. and she passed out and shook uncontrollably for about 20 minutes. She had the same reaction for the following four weeks. Now she is given preparatory meds be-fore the phlebotomy. Dealing with the PCT has continued to be difficult but Amy deals with it like she does all else...HEAD ON. Amy was also diagnosed with multiple sclerosis which complicated her health. Despite both PCT and MS, Amy decided to help others who had PCT and all porphyrias, so she promptly joined the APF and volunteered her time and energy and is now on our staff. Her experience as a former teacher and school owner has been invaluable to the APF.

Thank You , Amy!

— Pharma Friends — GEORGE MENSING

The APF has initiated a column about the people in the pharmaceutical industry that help develop and provide treatments for different types of porphyria. Collaborating with these new friends has been a wonderful experience as they have the same goals as the APF .... to enhance your health and lessen the suffering.

George writes, "My name is George Mensing, Director of Clinical Operations at Disc Medicine. I've had the pleasure of working at Disc for almost 3 and half years now and in clinical trials for 15 years, mostly in rare diseases. I started my career in research with the goal of contributing to science in the pursuit of bettering the lives of people, first those suffering from chronic pain, then transitioned to rare hematologic diseases, which brought me to Disc. It was here that I first learned of EPP and the incredible stories of patients' resilience in the face of such a painful, unseen, and underserved disease. Bitopertin and its mechanism of action struck me as a promising treatment that could help better alleviate the sensitivity to sun, and potentially ad-dress the underlying cause of EPP and XLP and its impact on the hepatobiliary system. It has been a great honor working at Disc surrounded by passionate experts that are focused on making patients' lives better. Leading the Phase 2 trials and overseeing the execution of APOLLO, our Phase 3 trial, working with advocacy groups like the APF, and EPP physicians and experts, has been the highlight of my career.

Outside of my day job, I am a full-time dad to our two little ones, Will and Evie, ages 5 and 3. My wife Katie and I live a few miles north of Boston. Our weekends are filled with swim lessons, gymnastics, dance, tee-ball, and nonstop drawing. We are looking forward to putting the kids' new swim skills into action this summer and have mapped out 8 beaches, ponds, lakes, and swimming holes across New England (Will literally drew a map). I've recently gotten into pickleball, and I think it's fair to say that it is an obsession at this point.

NEED SUPPORT AND WANT TO GIVE SUPPORT TO FELLOW PORPHYRIA PATIENTS. JOIN OUR PORPHYRIA PARTNERS PROGRAM.

CALL 866-APF-3635 for details!

ADVOCACY

The Photodermatology Society... Advancing Knowledge On Light-Related Disorders

The field of photodermatology plays a critical role in the diagnosis and management of various skin disorders affected by light, including cutaneous porphyrias. The Photodermatology Society, under the leadership of Dr. Elizabeth Buzney, President and member of the APF Scientific Advisory Board, is committed to enhancing un-derstanding of these complex conditions. This society is particularly important to patients suffering from the cutaneous porphyrias, a group of disorders characterized by sensitivity to light, leading to painful skin reac-tions and other complications.

Recently, the Photodermatology Society held a significant event—the Annual Scientific Meeting—prior to the American Academy of Dermatology's annual convention. This day-long gathering brought together experts and professionals to further explore the role of light in health and disease. One of the key highlights of the meeting was the keynote address delivered by Dr. Robert Sarkany, a leading porphyria expert from the UK. His presentation on the porphyrias was both informative and enlightening, offering fresh insights into the challenges faced by individuals with these light-sensitive conditions.

Dr. Buzney's leadership and tireless efforts in the field of photodermatology have been instrumental in ad-vancing the understanding of these conditions. As a clinician, she provides specialized care to patients with Erythropoietic Protoporphyria (EPP) and X-linked Protoporphyria (XLP) at Brigham and Women's Hospital in Boston. Her work not only provides vital treatment to patients but also contributes to the growing body of knowledge surrounding these rare and often misunderstood diseases.

The Photodermatology Society's commitment to expanding knowledge and fostering collaboration among ex-perts in the field is crucial for the development of better diagnostic methods, treatments, and preventive measures for those affected by light-induced skin disorders. Thanks to Dr. Buzney's dedication and leadership, the Society continues to make strides in improving the lives of individuals living with cutaneous porphy-rias and related conditions. Her efforts are helping to shed light on the complexities of light-induced disor-ders and bring hope to many who are affected by these challenging conditions. See http://photomedicine.org/

CLINUVEL Pharmaceuticals Shines at AAD Convention with Photomedicine Pavilion

Recently, CLINUVEL Pharmaceuticals hosted the most dynamic and innovative exhibit at the American Academy of Dermatology (AAD) Convention, held in Orlando, Florida. Their Photomedicine Pavilion united science and art in a groundbreaking display, captivating attendees and highlighting CLINUVEL's pioneering work in the field of photomedicine.

The exhibit was a visual feast, featuring a variety of dynamic shows, patient stories, media programs, and educational content from past years. One of the most unique aspects of the pavilion was a dedicated room that paid tribute to the trailblazers of photomedicine, with photos and profiles of key figures like Dr. Elisabeth Minder and Desiree Lyon (*photo*), whose groundbreaking efforts in the development and education of pho-tomedicine have shaped the industry.

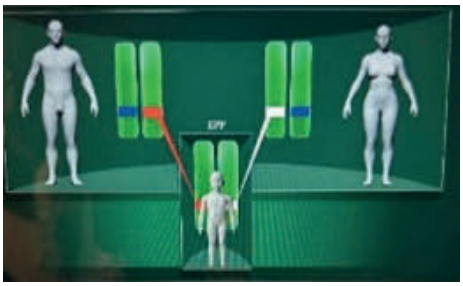
Nicole Castellano, APF Director, shared the ongoing work of the APF and the importance of patient advocacy with many of the attendees. She also expounded SCENESSE, Clinuvel's treatment for EPP and XLP. As the only FDA approved treatment for EPP and XLP with a stellar 20-year safety record, patients describe SCE-NESSE as life changing, and revolutionary. If you are interested in gaining access to SCENESSE or the new variegate porphyria VP trials, contact the APF at 866-APF-3635. RE VP trials, See:

https://www.clinuvel.com/2024/12/health-canada-validates-scenesse-new-drug-submission-20241223/

Watch this outstanding Clinuvel video explaining EPP,

https://www.clinuvel.com/pharmaceuticals/disease-entities/erythropoietic-protoporphyria-epp/

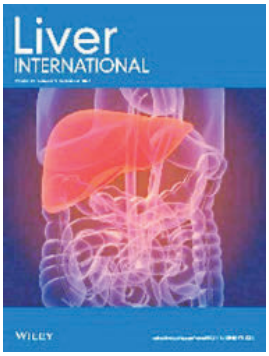
Thanks so Clinuvel for the graphic



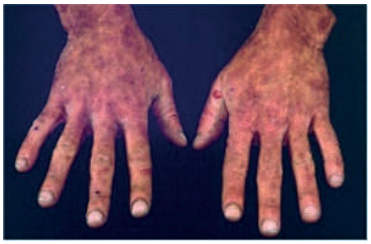
CRISPR

Much of the upcoming research in the world today involves CRISPR (Clustered Regularly Inter-spaced Short Palindromic Repeats). CRISPR is a gene editing tool that allows researchers to modify DNA in living organisms and is based on a defense system that was found in bacteria and other microbes. Basically, CRISPR is a system that can target and edit specific DNA sequences. Since it is an easy process and very precise, it can modify hundreds of genes at once. Thus, CRISPR can correct mutations in the human genome to treat genetic diseases. We want to thank all of our members who participated in the recent CRISPR APF survey. It is our hope that one day, porphyria will be on the genetic treatment radar.

IMPORTANT READING for Physicians & Patients



Included in the Liver International is the publication on Long-term complications in acute porphyria by Dr. Elena Pischik with Mattias Lissing, Nicolas Pallet, Raili Kauppinen from the Department of Neurology, Consultative and Diagnostic Centre in St. Petersburg, Russia. Although the life expectancy of patients with acute porphyria has become comparable to that of the general population, long-term complications can lead to an inferior quality of life. Recurrent acute attacks can lead to chronic pain and fatigue, high blood pressure and chronic kidney disease. Moreover, patients have a high risk of primary liver cancer but not of other cancers. The best preventive treatment is to stop recurrent attacks with lifestyle is preventive preparations, such as hematin/Panhematin, givosiran /Givlaari or liver transplantation. Regular monitoring for blood pressure, kidney function and other cardiovascular risk factors, and liver cancer can be personalized by the patient's age, comorbidities and disease activity. Find the full publication in the Liver International Journal Magazine Volume 44, Issue 9, September 2024, Pages 2197-2207



Another Liver International publication is The clinical management of porphyria cutanea tarda: An update by R Sarkany Volume 44, September 2024. been significant advances in the understanding of the factors predisposing to the disease, and of its wider health impacts. Porphyria cutanea tarda (PCT) is the most common type of porphyria, which is either inherited or occurs secondary to an underlying internal disorder. Dr. Sarkany elucidates the significant advances in the understanding of the factors predisposing to the disease, and of its wider health impacts. This review aims to help the clinician to diagnose and manage pa-tients with PCT, with an emphasis on the impact of recent advances on clinical management. See https://onlinelibrary.wiley.com/doi/10.1111/liv.15980.

Dovepress

Obstacles to Early Diagnosis of Acute Hepatic Porphyria: Current Perspectives on Improving Early Diagnosis and Clinical Management by Drs. M. Thapar and A. Singh present the many pitfalls and challenges in the diagnosis of acute porphyrias in this Dovepress publication. The obstacles to diagnosis include inappropriate urine testing for porphyrins only as opposed to urinary testing for the accumulation of PBG/porphobilinogen and ALA/amino levulenic acid and creatinine levels, inadequate sample handling, and ordering genetic testing as the initial diagnostic test. These are other challenges are noted in the following link https://www.dovepress.com/obstacles-to-early-diagnosis-of-acute-hepatic-porphyria-current-perspe-peer-reviewed-fulltext-article-CEG

Congenital erythropoietic porphyria five years observation with standard treatment: a case report by M. Kamalyan and M Mohammadi concerns a 22-year-old Armenian man suffering from the typical symptoms of CEP, photosensitivity, excessive hair growth, mutilation, and pink urine discoloration. After five years of tracking the condition, it was revealed symptoms worsened despite preventative measures. Thus, standard recommendations did not lessen the patient's deteriorating conditions. A cure with an al-logeneic haematopoietic stem cell https://academic.oup.com/omcr/article/2024/1/omad151/7590470.

SCIENCE DIGEST Canadian guidance for diagnosis and management of acute hepatic porphy-rias by Dr. David Colantonio in Clinical Biochemistry Volumes 131-132, October 2024 110792 addresses the challenges, complications and delays in diagnosis and management of the acute porphyrias. The aim is to improve awareness and medical outcomes of acute porphyrias by making recommendations about diagnosis, monitoring, and treatment in Canada.

NEED ASSISTANCE?
CONTACT THE APF AT
866-APF-3635

What's New?

Check out www.PorphyriaFoundation.org

The information contained on the APF website or newsletter is provided for 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 consult with personal physician or local treatment center before pursuing any course of treatment.

Donate to the APF

Donate on our website 24 hours a day, 7 days a week. Thank you!

THANK YOU!

The APF does not receive government funding. Your contributions help educate physicians and patients with life-saving information about the porphyrias. Donations are tax deductible. Become an APF member today!

UPDATED CONTACT INFORMATION?

Contact 866-APF-3635 or general@porphyriafoundation.org.

AMERICAN PORPHYRIA FOUNDATION
6605 33rd St E, Suite C Sarasota, Florida 34243

Address Service Requested

Non-Profit
U.S. Postage
PAID
Suburban, MD
Permit No. 264