AMERICAN PORPHYRIA FOUNDATION

WINTER 2025 **NEWSLETTER**

IN THIS ISSUE

MERRY CHRISTMAS AND HAPPY HOLIDAYS FROM ALL OF US AT THE AMERICAN PORPHYRIA FOUNDATION!

As we celebrate this season of gratitude and hope, we extend our heartfelt thanks to our porphyria community—patients, families, caretakers, physicians and industry partners—for your ongoing support, courage and compassion. May your holidays be filled with peace, joy, and good health, and may the new year bring continued progress and brighter days for all our friends living with porphyria.

FONDEST GREETINGS,
Tom, Jessica, Pax, Debra, Andrew, Nadene, Nicole, Paul, Warren, Ron, Diana, and Desiree

Porphyria Documentary Coming to Netflix

We are proud to announce a collaboration with Myth Entertainment and Netflix on the soon to be released documentary, *It's All in Your Head*. This powerful film will shed light on the lived experiences of individuals affected by porphyria, a group of rare genetic disorders often misunderstood or misdiagnosed for years.

"It's all in your head" is a poignant reflection of what many porphyria patients endure during their diagnostic journey. Because porphyria presents a wide range of complex and painful symptoms, healthcare professionals unfamiliar with the disease often attribute these experiences to psychological causes rather than physiological ones. This misconception leaves many patients without answers and to struggle for validation and appropriate care. Our Founder and former Director Desiree Lyon says, "Patients with acute porphyria, in particular, have faced immense challenges due to delayed or incorrect diagnoses. This documentary brings much-needed awareness to their stories and to the urgent need for education among healthcare professionals."

Here at the APF, we proudly assisted in creating this important documentary by connecting patients and experts to contribute insights and experiences that ensure accuracy and authenticity. Stay tuned to our e-news, website, and social media for updates on when *It's All in Your Head* airs in Spring 2026.

BE FEATURED IN THE CREDIT SEQUENCE

Together, we can raise awareness and change the narrative for those living with porphyria. Donate $50 or more to be featured in the *It's All in Your Head* documentary credits as a sponsor. Corporate and major donors will receive special recognition. Become a sponsor today by donating online or sending a check to the American Porphyria Foundation at 6605 33rd St. E Suite C, Sarasota, FL 34243.

For online donations, write "sponsor" in the "in honor" field.

Presidential Award Honoree: Dr. Roy Smith

The APF Presidential Award is bestowed upon people who dedicate their lives to porphyria advocacy. Dr. Roy Smith is a professor of hematology at the University of Pittsburgh's Vascular Medicine Institute and widely recognized among the world's foremost authorities on porphyria. His research illuminates the most complex aspects of heme biosynthesis and porphyrin metabolism, providing critical insights into the genetic and biochemical roots of the porphyrias. His work has strengthened diagnostic testing, improved clinical trial design, and advanced the natural history studies that underpin modern treatment approaches. Through decades of rigorous science, he has laid a foundation on which new therapies and patient resources continue to be built. Dr. Smith has not only served patients as a clinician and researcher, he also is willing to advance porphyria awareness in any way possible from Zoom calls, podcasts, documentary participation to porphyria publications. Few individuals embody the balance of scientific brilliance and deep compassion for patients as Dr. Smith.

Dr. Roy Smith, Scientist, Clinician, and Champion for Porphyria Patients, is honored with the 2025 APF Presidential Award.

We're proud to recognize and honor Dr. Smith as a guiding light for patients, families, and the scientific community with this year's APF Presidential Award.

EPP School Guide Launch

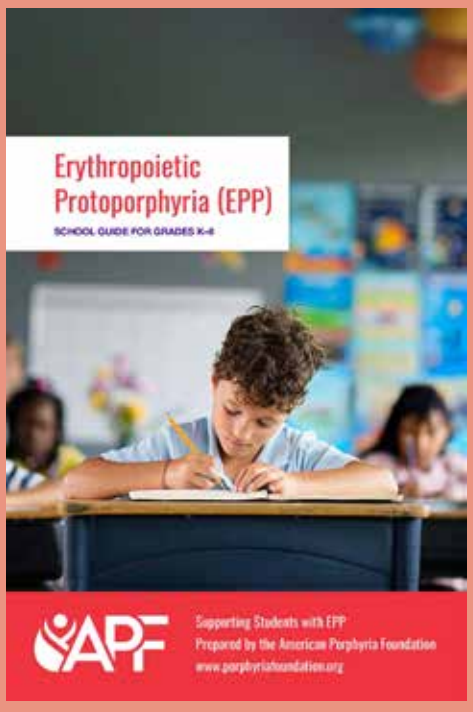
Kids attending school jump substantial hurdles to stay safe and happy when they live with Erythropoietic Protoporphyria (EPP) or X-linked protoporphyria (XLP). Here at the APF, we've been collaborating with folks who live with EPP, doctors and their families to create a school guide. Full of recommendations, accommodations and suggestions, the guide will help educate and facilitate particular conversations among families, teachers, administrations and classmates. Contact 866-APF-3635 for a FREE copy!

REQUEST A FREE EPP SCHOOL GUIDE!

Contact us at 866-APF-3635 or **general@porphyriafoundation.org.**

The EPP School Guide is one of many projects we'll be launching early next year as part of our "Shade Squad" program, created especially for kids with EPP or XLP. Stay tuned for Shade Squad comics and coloring books filled with sun-safe puzzles, games, badges and other meaningful activities for kids with EPP. Plus, the program will offer an opportunity to connect families with a child impacted by EPP and XLP. Our global sister organizations will receive copies of the school guide as well, to distribute to families around the world.

We couldn't have written this guide without the help of volunteers, George Hodder and Steve Ferry, who serve on our Member Advisory Board (MAB), and have EPP. They channeled frustrations and wins from their own school days to create a scenario of best practices that served as the basis for this guide. Their participation and leadership represents just one example of why the MAB is paramount to serving the porphyria patient community.



Thanks to Clinuvel who was instrumental in turning our ideas for an EPP School Guide into a professional booklet we'll distribute worldwide. Thanks also to Disc Medicine and Mitsubishi Tanabe USA for your support with this project.

Pharma Friends

In each newsletter, we spotlight a friend from the pharmaceutical community — people who dedicate their careers to advancing treatments that improve and save lives. Our "Pharma Friends" are everyday individuals who care deeply about their families, their work, and the patients they serve.

Samantha Hurley of Alnylam Pharmaceuticals and her daughter, Quinn.

I'M SAM HURLEY, I WORK IN PATIENT ADVOCACY & ENGAGEMENT AT ALNYLAM PHARMACEUTICALS.

When my daughter was born last year with a mix of rare and non-rare conditions, my professional mission became deeply personal. I now split time between the children's hospital and Alnylam, partnering with patient advocacy groups like the APF to support patients and care partners as they navigate rare diseases. I feel so lucky to work for a company helping patients with acute hepatic porphyria and enjoy helping patients get diagnosed, access the right treatment(s) for them, and find positivity and support along their path.

Outside of advocacy work, I'm training to become a Jazzercise instructor and enjoy life with my husband, daughter Quinn, and our two dogs, Waffles and Barley, in Austin, Texas. I love baking, paint-by-numbers, and the outdoors.

New Scientific Advisory Board Members

The Scientific Advisory Board is essential to our mission. These doctors and scientists are renowned in their fields, and provide oversight for our patient and physician education programs.

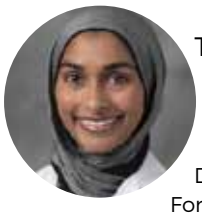
We are proud and honored to welcome these new members to our Scientific Advisory Board!

JONATHAN ARAMBULA, PHD

is one of the top porphyrin researchers in the world and co-founder of InnovoTEX, a biotech company that's redefining cancer treatment. With a background spanning academic research and executive leadership, Dr. Arambula has built a career developing targeted therapies that bridge the gap between science and real-world patient impact. He also lives with EPP. Listen to Dr. Arambula explain the role of porphyrins on our *Rarely Discussed* podcast.

ROBERT SARKANY, MD

is one of the world's esteemed EPP experts and researchers. He is a senior consultant dermatologist at St John's Institute of Dermatology. He's an international authority in the area of sunlight allergies and ultraviolet light therapy. Dr Sarkany began his porphyria career researching in Cambridge, where he was awarded a Doctorate in Medicine for his research into sunlight sensitivity. He also researched the molecular biology of porphyria in Heidelberg, Germany.

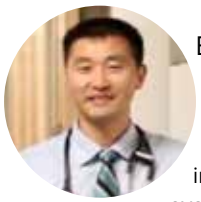


TASNEEM F. MOHAMMAD, MD

is a senior staff physician in the Department of Dermatology and at Henry Ford Hospital in Detroit. She's also the Director of the Skin Discoloration Clinic and the EPP Clinic, where she provides specialized care for patients with complex pigmentary disorders and photodermatoses, like the porphyrias. She is involved in clinical trials focused on EPP, photoprotection, and pigmentary disorders, and her work bridges clinical care and research to improve outcomes for patients with rare and challenging skin conditions.

JEHANZEB RIAZ, MD

is a medical oncologist with Oklahoma University Health Stephenson Cancer Center in Tulsa, who has now taken on porphyria as a new specialty. Dr. Riaz says, "I focus on helping people understand their disease, challenges and treatment options to maximize their chance to prolong survival and improve quality of life. It is very important that the patient is treated respectfully and ultimately, is given a feeling of empowerment." Dr. Riaz treats all common cancers such as breast, lung, colon, and prostate, and is fluent in Hindi and Urdu.



BO ZHAO, MD, PHD

is a board-certified hematologist at Virginia Oncology Associates in Williamsburg, with over 20 years of expertise in clinical medicine, hematologic disorders, and translational biomedical research. Dr. Zhao has a specialized interest in the diagnosis and management of porphyrias. His clinical work integrates the latest advances in medicine to offer diagnostic strategies and individualized care for patients with acute and cutaneous forms.

Breakthroughs in CEP Research

Dr. Oscar Millet from Spain is changing the hopes and dreams of congenital erythropoietic porphyria (CEP) patients worldwide. He led a team in developing a drug called "ciclopirox" and navigated through complicated regulatory hoops to receive orphan drug status from the FDA and European Medicines Agency in 2022.

After confirming the ciclopirox's safety in Phase I trials, his biggest shock was the bureaucracy. "The paperwork is probably more than 10,000 pages sent by the FDA," he says. Fortunately, Dr Millet's years of research is headed into clinical trials in the United States. This news brings hope for a CEP treatment to the forefront and a step closer to reality.

Dr. Óscar Millet has been awarded for scientific excellence by the Royal Spanish Society of Chemistry. and the 2025 Research Excellence Award by the Royal Spanish Society of Chemistry in recognition of his outstanding scientific career. He has driven key initiatives and made significant advances in the study of rare diseases. With over 140 scientific publications and multiple awards, his research has had a notable international impact. Thanks to Dr. Millet and his dedicated team for the wonderful, life changing work for people with CEP across the globe.

Read more about Dr. Millet and CEP breakthroughs.

Over years of research, CEP Patient and Spanish Porphyria Association Director Fide Mirón has closely collaborated with Dr. Millet's team as a research participant.

FDA to Review Bitopertin within Two Months

Disc Medicine submitted a New Drug Application to the U.S. FDA, seeking accelerated approval and priority review for Bitopertin to treat EPP and XLP in patients aged 12 and older. They received the Commissioner's National Priority Voucher, part of a new FDA program, which can shorten drug review times to within 60 days.

disc medicine

The Youngest Patient on SCENESSE®

A nine-year old child in Switzerland has been treated with SCENESSE®. She is the youngest person to receive this revolutionary treatment for EPP, and our community is buzzing with this news! We are pleased to report she is doing very well, and we'll keep you updated as we know more.

REQUEST A FREE DOCTOR PACKET!

Contact the APF at 866-APF-3635 or **general@porphyriafoundation.org.**

WE DO **NOT** SHARE INFORMATION ABOUT MEMBERS WITHOUT THEIR PERMISSION.
WE DO **NOT** SHARE MEMBERS' NAMES TO PHYSICIANS WITHOUT THEIR PERMISSION.

New Publication Highlights Barriers to AIP Management

The Recordati Rare Disease Metabolic Team is delighted to announce *Acute Intermittent Porphyria: An Overlooked but Treatable Inherited Neuropathy* by The Clinical Neurological Society of America. This published research delves into the burden and impact of acute intermittent porphyria (AIP) and barriers to its diagnosis and management. Distinguished contributors include porphyria experts: Mohamed Kazamel, MD (Chair), Mayo Clinic Arizona; Richard Andrews, MD, CHI Health Omaha; Manisha Balwani, MD, MS, FACMG, Mount Sinai; Eddy Lang, MD, University of Alberta Calgary; Rachana K. Gandhi Mehta, MD, MBBS, Wake Forest University; and Bruce Wang, MD, University of California San Francisco.

Read the new publication about AIP!

Locate Panhematin Near You

We help people with acute porphyria access treatments every day, but finding Panhematin just got a whole lot easier. Use a new tool to identify hospitals and clinics that either have Panhematin available or are willing to order it. Go to **panhematin.com/find-panhematin** and type in your zipcode. Alternatively, you can call 1-866-738-0800.

Participate in New Research

If you have AIP, VP or HCP and haven't received treatment for an attack you've endured in the last two years, you may be eligible for a new research study. Participants will be interviewed for 90 minutes and compensated $240. Contact 866-APF-3635 or email **general@porphyriafoundation.org.**

Warning for Acute Patients Considering Hormone Therapies

United Kingdom Porphyria Medicines Information Service recently shared important guidance on the use of hormone replacement therapy (HRT) for those with acute porphyrias, including AIP, variegate porphyria (VP), and hereditary coproporphyria (HCP).

Some people with acute porphyria are at risk of severe acute attacks, often starting with abdominal, back or thigh pain, alongside symptoms such as nausea, vomiting and constipation. Attacks can be triggered by many factors including medications, alcohol, fasting, illness, stress as well as hormones, particularly progesterone.

Women are at increased risk if they have a history of acute attacks, are in their late teens to early forties, or are pre or peri-menopausal. During menopause, estrogen and progesterone levels naturally fall. HRT is used to replace these hormones to relieve symptoms. Both hormones can trigger attacks, with estrogen thought to be less risky than progesterone.

If HRT is needed:

- Consider estrogen-only HRT when possible
- Try topical forms (creams or gels), which are generally safer than oral tablets
- Avoid hormonal implants or injections, as these cannot be removed if an attack starts

If prescribed HRT:

- Remind your healthcare provider repeatedly that you have acute porphyria
- Use the lowest dose possible to manage symptoms
- Stop HRT and contact your porphyria specialist immediately if you develop abdominal pain or other attack symptoms

Please consult your doctor as this information is not specific to each patient. Where possible, menopause symptoms can be managed individually, taking into consideration the unsafe drug database on **porphyriafoundation.org.**

Podcast Episodes on Website

It's now easier than ever to watch our *Rarely Discussed* podcast. Visit **porphyriafoundation.org/podcast** to find all past and present episodes.

APF Member Keshia Johnson recently appeared as a guest and you can now listen to her inspiring story on our website. Are you interested in being featured on the podcast or know someone who is? Call us at 866-APF-3635 or email **general@porphyria.org.** Our producer and host, Andrew McManoman, will guide you through the easy process of telling your story.

Doctors also make great guests, as they can share the medical education pertinent to porphyria diagnosis and treatment. Ask yours about it!

Watch Keshia Johnson on a recent episode of *Rarely Discussed*.

Porphyria and Rare Disease Assistance

Every day, we provide and connect people impacted by the porphyrias with resources and assistance. Living with a rare disease is expense and complicated. But here are some programs available to help with medication costs, financial assistance with insurance premiums and co-pays, disability benefits information and travel assistance for clinical trials or consultation with disease specialists. Call 866-APF-3635 with specific questions.

CLINUVEL'S PATIENT ASSISTANCE

This program helps patients with EPP access SCENESSE® (afamelanotide) treatment, and is open through December 15, 2025. If you are eligible and would like assistance, please contact 866-APF-3635 or **general@porphyriafoundation.org**.
We are committed to helping EPP patients receive an FDA-approved treatment by educating patients and their medical team and assisting them with gaining access to treatment.

THE ASSISTANCE FUND

The Assistance Fund recently announced the launch of its new financial assistance program for people living with porphyria. The Assistance Fund is an independent charitable patient assistance foundation that helps patients and families facing high medical out-of-pocket costs, such as copayments, health insurance premiums, and incidental medical expenses.
Visit **tafcares.org.**

HEALTHWELL FOUNDATION

When health insurance is not enough, the HealthWell Foundation fills the gap by assisting with copays, premiums, deductibles and out-of-pocket expenses for essential treatments and medications. HealthWell Foundation is a nationally recognized, independent non-profit organization that has served as a safety net across over 80 disease areas for more than 615,000 underinsured patients by providing access to life-changing medical treatments. Visit **healthwellfoundation.org.**

SOCIAL SECURITY DISABILITY

If you have been diagnosed with porphyria and it has significantly affected your life, you may be eligible for Social Security Disability benefits or Supplemental Security Income. These benefits can help toward paying medical treatment bills or other expenses. If you need a letter for Social Security Disability about your type of porphyria, we will assist you. Please email us at **general@porphyriafoundation.org.**

AGENT ORANGE VA BENEFITS

Agent Orange exposure during the Vietnam War has been linked to PCT. We help veterans exposed to Agent Orange and then diagnosed with Porphyria Cutanea Tarda (PCT) receive financial assistance and medical help. The US Department of Veterans Affairs (VA) presumes the condition is related to their service if it develops within a year of exposure and is at least 10% disabling.
Call us at 866-APF-3635 and read more information below.

MORE ON AGENT ORANGE

The VA presumes Agent Orange exposure in certain locations and time frames to make it easier for veterans to receive compensation for related health conditions, including PCT. If a veteran served in Vietnam, along the Korean DMZ, or in certain locations in Thailand during the Vietnam War era and has been diagnosed with a condition recognized as linked to Agent Orange, they may be eligible because the VA presumes the condition is service-connected. Visit the VA website for more information.

Autumn Patient Get Togethers

This Fall, members of our porphyria community gathered in Venice, Fla. and Philadelphia, Penn. The crowd assembled and immediately began sharing their lives. That is the wonderful part of in person meetings. Although social media provides the opportunity to chat online as a group, it does not provide the level of connection that a face-to-face meeting allows.

We know of friendships that have lasted over 30 years, when the APF held our first patient meetings. The size of the meeting is immaterial, it is the joy of being able to build relationships with others in your unique situation. Sharing your rare aspects of your porphyria lives with someone who's also been through it is a life-altering experience. All the porphyrias are rare, some rarer than others, so the chance of meeting another person with your same disease is not likely without meet-ups like these.

A big thanks to Amy and Craig Chapman, and Gudrun Debes for hosting. Thanks also to Platinum Sponsor Alnylam Pharmaceutical, and Silver Sponsors Recordati Rare Disease and Disc Medicine for helping us bring our community together.

APF Members Gudrun Debes (left) and George Hodder (right) met at the Philadelphia patient meeting in November.

Meet Friends with Porphyria

Bring your family and friends and enjoy meeting new porphyria friends while learning about the porphyrias. Watch our social media and e-news for upcoming meet-ups in Atlanta, Kansas City, Oklahoma City, Chicago, Dallas, San Francisco, Los Angeles, and/or San Diego and Seattle.

These meetings are made possible by APF members, who are willing to host in their cities and towns. Call us at 866-APF-3635 if you'd like to set up a patient gathering near you! See your city listed? We're always looking for volunteers to help with the events.

Monthly Virtual Hang Outs

Stay up to date on book picks! Join with **general@porphyriafoundation.org.**

Love crafting? Here's your space to create among porphyria friends. Email **general@porphyriafoundation.org** for details on the next meet-up!

APF friends often give donations in memory or honor of their dear loved ones. We are grateful for each of them.

IN MEMORY

Daniel Pudlicki
Matthew E. Cole
Carl Rusnak
Ellane Heflin
Ruthie Loomis
Sharon Stapleton
Darlene Bishop
Sylvester John Berdak

IN HONOR

Aaron Pudlicki
Giner Davis
Tasha & Kalel Alicea
Eliza Maria Martinez-Ureste

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 a consult with personal physician or local treatment center before pursuing any course of treatment.

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

THANK YOU!

Why donate to the APF?

We've put patients first for 40+ years. Staff work nights and weekends helping people worldwide find answers for painful symptoms.

Your donation helps us provide doctor packets for newly diagnosed, add to a growing database of 6,000 treating physicians worldwide, support physician education, and more!

Every donation is tax deductible and incredibly appreciated. The APF does not receive government funding.

NEED TO UPDATE YOUR CONTACT INFORMATION?

Contact 866-APF-3635 or **general@PorphyriaFoundation.org.**

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