AMERICAN PORPHYRIA FOUNDATION

SUMMER 2024 **NEWSLETTER**

IN THIS ISSUE

Thanks for Thinking Porphyria!

National Porphyria Week was April 13th — April 20th, and we had the country thinking porphyria!

DR. ROY E. SMITH

Thank you, Dr. Smith! Back by popular demand, Dr. Roy E. Smith, a hematologist and professor at the University of Pennsylvania School of Medicine, shared porphyria expertise on the NPAW kick-off zoom for the acute porphyrias (AHPs), PCT and CEP.

Thank you for clicking to support the APF, and thank you, Recordati! Our annual click campaign resulted in a $5,000 donation from Recordati Rare Disease for education and awareness programming. Recordati donated $5 for every click on the APF home page.

Thank you, Mitsubishi! Mitsubishi Tanabe Pharma America sponsored many NPAW activities in recent years. Mitsubishi is researching a new EPP drug, Dersilmelagon (MT-7117), and supports activities to help patients' emotional well-being and quality of life.

NPAW DAY-BY-DAY

Satisfy the Soul Sunday

Motivational Monday

Talk Tuesday premiered *Rarely Discussed,* APF's podcast with Director Nicole Castellano and Member Andrew McManamon. Find it on your favorite podcast application.

Wear A Hat Wednesday was the day for EPP kids to wear hats to school.

Tell Your Story Thursday

Film Friday featured *Out of the Darkness, Into the Light: Chris's Story, Part 1,* a documentary about the EPP experience of Member Chris. Watch on social, and stay tuned for part two.

Purple for Porphyria Saturday closed out NPAW with purple clothes, hats, glasses, hair and even feather boas!

NPAW IS ABOUT WHAT **YOU'RE** DOING FOR AWARENESS

Mayra and her son-in-law, Carlos Lopez, partnered with his San Diego-based employer, Truly Nolen of America, Inc., to host a porphyria awareness booth. Truly Nolen is a pest control company that supports causes close to their employees' hearts, and pledges to spread porphyria awareness at monthly events.

"It took five painful years, paralysis, and 24 hours to live before I was diagnosed with porphyria," Mayra said. "During NPAW we can teach people to 'think porphyria.'"

Member Mayra Martinez and son in law Carlos spreading awareness during NPAW.

DR. ELIZABETH BUZNEY

APF Scientific Advisory Board Member, Dr. Elizabeth Buzney, led a Zoom on erythropoietic protoporphyria (EPP). She is president of the Photodermatology Society, director of dermatology at Harvard Medical School and Brigham and Women's Hospital. **Watch on the APF YouTube channel.**

MEMBER STORIES

Easy as Wearing a T-Shirt

Recently, APF Global Patient Engagement Coordinator Pax Cumming (above), was stopped by a man on the street. “Porphyria!” he exclaimed, pointing at Pax’s t-shirt. “Wow, that’s rare.” Turns out, he’s a hematologist in Austin, Texas and treats acute porphyria and porphyria cutanea tarda (PCT). He was blown away Pax worked for the APF, and now accepts referrals in his area.

Last month, APF Director Nicole Castellano (above), wore her “Think Porphyria” sweatshirt as she traveled. Three people stopped her to ask about porphyria at the airport, and it provided her a great opportunity to spread awareness. Later that day when she arrived at her hotel, a man on the elevator noticed too. “Of course I know what porphyria is,” he explained. “I’m a hematologist!”

Check out the APF online store for your own advocacy gear!

A Life-Saving Collaboration

Moustapha is in his first year of a master’s program in Mathematics and Applications at the University of Nouakchott in Mauritania. He is alive today thanks to the partnership of family, doctors, industry, and patient organizations.

“I contracted mild flu, took antibiotics, and subsequently experienced severe stomach pains with continuous vomiting. I was rushed to the hospital, where I received medications that intensified my pain, leading to a coma,” Moustapha said.

His mother consulted Dr. Sejad, who identified porphyria in their family. His brother, Sidi, contacted Dr. Laurent Gouya in Paris, who reached out to Recordati and obtained emergency Normasang (Panhematin). After four days of treatment, Moustapha was moved out of the intensive care unit.

Dr. Laurent Gouya was instrumental in helping Moustapha access Normosang for an AHP attack

CHEERS & TEARS

CHEERS

Long-time Member Nathan Wayne Carr welcomed a granddaughter!

Kate Hudson, daughter of board members Dr. Judith and Warren Hudson, received her degree in Bio-Medical Sciences from Texas A&M.

TEARS

KAMERON PATRICK KADINGER

Seven year-old Kam passed away on April 9, 2024. Kam had AIP and both his parents, Amanda (Jason) Jordan and Keith Kadinger, carry the genetic mutation. Many APF members participated in the Kure4Kam campaign to help find treatment for his devastating illness. The APF facilitated a meeting of experts nationally and internationally to discuss his case and try to provide help. We express our sympathy to the many people who loved Kam.

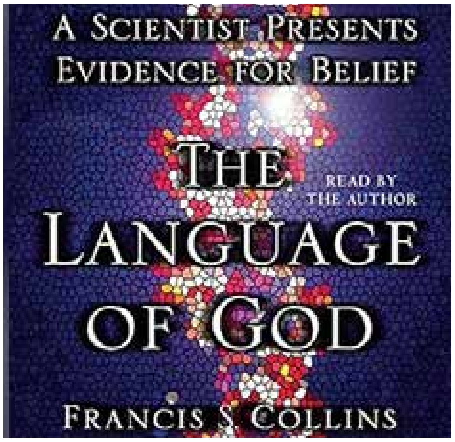
ERIC LIFSCHITZ

One the APF’s oldest members, Eric Lifschitz, died on May 6, 2024. Over 43 years with AIP, Eric used hospitalizations, ICU, and clinic visits as opportunities to educate medical staff. Eric endured more than most, surviving critical AIP attacks, liver failure, numerous liver ablations, blood transfusions, severe infections and constant burning pain. He praised his doctor, Dr. Neil Nagovski, for saving his life. Thanks to Eric for his continued support of the APF and his campaign to heighten porphyria awareness. Eric was loved deeply, and will be missed.

Meet APF President, Paul Stickler

Paul Stickler discovered his desire to help people with porphyria when his role at a pharmaceutical company representing Panhematin, the first orphan drug approved to treat acute porphyria, led him to the APF. Paul is a senior biotech executive specializing in the commercialization of rare and orphan diseases, as well as gene and cell therapies. But Paul's impact extends far beyond his professional responsibilities, demonstrating a personal commitment to improving the lives of those affected by rare diseases. The APF recognized his invaluable contribution by inviting him to join the Board of Trustees, where he recently became President. His transition not only highlights Paul's leadership qualities but also underscores the trust and confidence placed in him by the porphyria and rare disease communities.

APF Board President, Paul Stickler, is dedicated to improving the lives of people living with porphyria.



Dr. Francis S. Collins wrote *The Language of God: A Scientist Presents Evidence for Belief.*

DNA: The Language of God

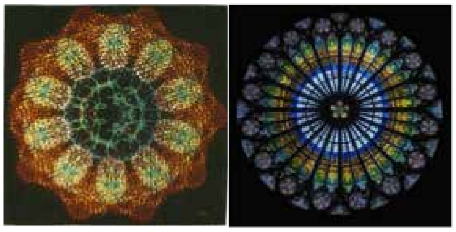
Dr. Francis S. Collins, M.D., Ph.D., is the former director of the National Institutes of Health (NIH). As the longest-serving NIH director — spanning 12 years and three presidencies — he oversaw worldwide biomedical research, from basic to clinical research. Dr. Collins is a physician-geneticist noted for his landmark discoveries of disease genes and his leadership of the international Human Genome Project, which culminated in April 2003 with the completion of a finished sequence of the human DNA instruction book. Dr. Collins has authored several books, including his bestselling 2006 title on DNA and his faith journey, *The Language of God: A Scientist Presents Evidence for Belief.*

Porphyria Media Coverage

Remember the TV series *House*? *House* was one of many series that featured porphyria, thanks to the APF's media program. Roland Woerner and retired APF Director Desiree Lyon started the initiative 35 years ago by submitting storyline ideas to broadcast networks, television studios, and publishing houses. Find a list of porphyria episodes and published stories under "media coverage" on the APF website.

Centers of Excellence

Our Centers of Excellence program will enable patients and physicians to locate porphyria expertise without incurring long-distance travel expenses, or the frustration of locating a provider with porphyria expertise. By deeming practitioners and institutions "Centers of Excellence," the APF ensures proper education about these disorders and accurate information on treatment.



The first photo may look like the rose windows typical of French Medieval Gothic Cathedrals (shown in the second photo). Instead, the it is a cross-section view of DNA. If you look closely, you can see the DNA double helix. The similarity is remarkable, perhaps, because the inspiration came from the same source. For a fascinating explanation of the unique shape of DNA, see **goldennumber.net/dna.htm**. This beautiful identifier is what we each carry in our bodies, like a serial number, marking every human being as special and unique.

Act Fast to Prevent Damage

Ever stayed at home rather than seek medical help for a porphyria attack because you were too overwhelmed, or you had no one at home to care for the kids? Maybe you were in too much pain to go to the emergency room or you thought you could tough it out at home.

If you live with acute porphyria attacks (AIP, VP, HCP, and ADP), there are important reasons why you shouldn't wait to see a healthcare provider. Your nerves may be damaged no matter what level of pain you're experiencing. Nerve damage can be lifelong and extremely painful.

"Attacks that are not treated promptly can lead to severe neurological damage, require longer periods of recovery, or, in some cases, are fatal." (Source: Gouya L, Ventura P, Balwani M et al. *EXPLORE A prospective, multinational, natural history study of patients with acute hepatic porphyria with recurrent attacks.* Hepatology. 2020;71(5): 1546-1558.)

Why Does Nerve Damage Occur?

Acute porphyrias are caused by a deficiency in the activity of specific enzymes, which help to make heme. Heme is a molecule that carries oxygen through the bloodstream and throughout the body. The production of heme involves a number of steps, but without the specific enzymes, the heme production process is interrupted. The body can't keep up with demands for heme, and molecules especially harmful/toxic to your body's nervous system build-up.

Serious Consequences occur when attacks are not treated timely and can result in peripheral neuropathy, paresis, respiratory and bulbar paralysis, chronic neuropathic pain, and even death resulting from cardiac arrhythmia or paralysis.

APF Member SH says, "Unfortunately, I had many attacks. At first, I sought treatment immediately. As the hospital trips racked up, I began to avoid treatment because I was stuck in the ER for hours, and it was too depressing. Sadly, that was the worst thing I could have done because not getting treated contributed to my chronic nerve damage and neuropathic pain 24/7."

The APF will help inform your doctor the importance of acting fast. Doctor packets also contain forms for free DNA testing. Call 866-APF-3635.

VP Research

At present, CLINUVEL is conduciting a research study on SCENESSE® for Variegate Porphyria (VP) patients. The APF will be hosting a ZOOM in September to enlighten VP patients about the SCENESSE® study. Please watch the APF website, social media and Enews for a date announcement and to participate in the ZOOM presentation with two experts in field.

Pregnancy and Acute Porphyria

Since pregnancy is associated with increased levels of hormones such as progesterone; attacks could be precipitated by this hormonal swing. The following are two studies of pregnancy in acute porphyria.

British researchers Dr. Joanne Marsden and Dr. David Rees **studied the outcome of pregnancy in 15 women with acute porphyria.** Interestingly, they discovered the majority had minor porphyria complications. Others were severely affected by their pregnancies and needed Normosang/heme arginate for attacks. The Normosang was well tolerated by the mothers and babies. There have not been studies specific to the use of hemin in pregnancy, but it's used worldwide without ill effect.

Another study reported in the *Journal of Internal Medicine* women with AHP can develop pregnancy complications, including gestational diabetes, high blood pressure, a baby with low birth weight, and an Apgar score of 7 or less. This study did not indicate an increased risk of perinatal or pre-term birth.

An analysis determined whether risks were different in women with attacks or high PBG levels. Results showed women were divided into normal, mid-, and high- levels of urinary PBG, based on the highest urinary PBG level recorded during their lifetime. There were not enough records of people with VP or HCP for statistic analysis. The researchers found people with urinary PBG at a normal or mid-level range did not have a high risk for complications, but those with higher-range PBG were more at risk. All women with porphyria should follow disease guidelines, including proper nutrition, avoiding unsafe drugs, and monitoring carbohydrate intake and blood pressure.

ACUTE PORPHYRIA

Important: Givlaari/Givosarin and Low PBG

Recently, some on Givlaari/givosiran reported they could not gain access to Panhematin for breakthrough attacks because their PBG was low. Elevated PBG has previously been used in screening for attacks. Patients on Givlaari/givosiran may have fewer attacks, but these are often NOT accompanied by elevations in urine ALA, PBG, and porphyrins.

The Porphyria Consortium (PC) now advises the following: in a patient with known acute porphyria, the diagnosis of an attack is made on clinical grounds. Treatment can be initiated without waiting for urine PBG and porphyrin results.

If you are having difficulty getting Panhematin for breakthrough attacks, contact the APF at general@porphyriafoundation.org.

CUTANEOUS PORPHYRIA

Beware of "Melanotan" Products

SCENESSE® (afamelanotide), marketed by Clinuvel, is FDA-approved in the USA to increase pain-free light exposure in adult patients with a history of phototoxic reactions from EPP. Products sold online as "Melanotan" are unlicensed, unregulated chemicals that are wholly unrelated to CLINUVEL's development program or SCENESSE®. The sale of Melanotan for human use is illegal in most countries. Due to the unlicensed nature of the Melanotan products, they may pose a serious threat to human health, and are under no circumstances recommended.

Some EPP patients have reported that Melanotan is the same product as SCENESSE®. This is incorrect! SCENESSE® is a prescription containing the active substance afamelanotide, given subcutaneously by a trained healthcare professional. It is accepted by most insurers and has an excellent safety record. See Scenesse.com for more details.

Have EPP or XLP? Learn About a New Trial

Disc Medicine is conducting studies on Bitopertin, their new treatment for EPP and X-linked protoporphyria (XLP). looking at changes in protoporphyrin IX levels, safety, tolerability and measures of photosensitivity. A recent announcement about their AURORA study noted it was "a phase 2 study of bitopertin in patients with EPP. Treatment with bitopertin resulted in statistically significant reductions in PPIX, the primary endpoint, and significant improvements in the rate of phototoxic reactions with pain."

Mitsubishi Tanabe Pharma America, Inc. (MTPA) is investigating the efficacy, safety, and tolerability of dersimelagon (MT-7117), in adults and adolescents with EPP or XLP. Dersimelagon is an investigational treatment under study for its potential to increase the time in sunlight before patients experience symptoms such as burning, tingling, itching, stinging or fully develop a phototoxic reaction. Up to 150 patients with a confirmed diagnosis of EPP and XLP from the ages of 12-75 may qualify.

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

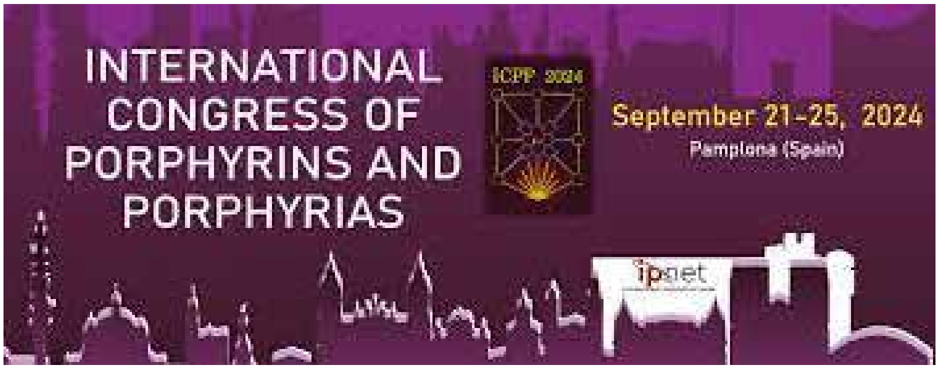
IN MEMORY

Gary Horn for **Sandra Horn**
George Rusnak Jr. for **Carol Rusnak**

IN HONOR

Eileen and Elayne for **Cecilia Decker**
Dr. Roy E. Smith for **the APF**
Dianne Fletcher for **Darlene Bishop**

International Porphyria Conference in Spain



Save the date! The 2024 International Congress of porphyrins and porphyrias (ICPP) will take place on September 21-25 in Pamplona, Spain. The ICPP brings together clinicians, researchers, scholars, patients, and their families for three days of networking and learning about the latest research in the porphyria field. The Spanish Team is preparing an exciting program to discuss the latest advances in our understanding of the natural history, management, and treatment of hepatic and erythropoietic porphyrias.

Accompanying social activities, include a tour of historical downtown, vineyards, and some of Spain's best-preserved medieval villages.

Patient Days on Saturday, September 21st and Sunday, September 22nd will include education and interactive workshops on topics such as the importance of genetic tests, diagnosis capabilities, available and innovative therapies, patient-centered care, the use of complementary medicines for healthy diets, mental health, an art workshop and updates from global and regional porphyria organizations. An art exhibit will feature pieces provided by attendees, as a creative way to raise awareness of its impact on patients and their families. Lunch on Sunday will provide a time for patients to connect

APF PODCAST GOES GLOBAL

APF's *Rarely Discussed* podcast will be broadcast from the ICPP in September. Guests will include worldwide experts, patients, caretakers, and industry. The podcast will be available globally and in the guests' languages.

An Unusual Porphyria Story in South Africa

Did you know that VP is the most prevalent genetic disease in South Africa? The history of porphyria began in 1652 when the Dutch East India Company agreed to bring a group of Dutch orphan women to the Cape "with an eye to marriage." One of these young women was Adriaantje Adriaanse van Rotterdam, who married a farmer named Gerrit Jansz van Deventer. The researchers don't know which one carried the VP mutation, but three hundred years later, we do know this couple started this estimated 30-40,000 VP patients in South Africa.

Porphyria research among the millions of Bantu people in South Africa began in the 1950s under Dr. H D. Barnes. Research was cut short after limited efforts to locate people with the disease. Now, the APF is pleased to collaborate with Aletta Longoria, who's worked with patients in South Africa for the past 20 years. For help or information, contact **general@porphyriafoundation.org.**

APF Director Educates

Nicole Castellano was invited to Alnylam Pharmaceuticals in Cambridge, Massachusetts to educate on acute porphyrias, unique challenges for patients, and her powerful personal story. Nicole also shared about the APF role in Global Porphyria Day, an event sponsored by Alnylam, and the upcoming International Porphyria Congress.

Nicole also was asked to present to the Swedish Hospital in Seattle as part of their program to educate residents and nurse practitioners.

Moving Porphyria Forward in Mexico

The new Vice President of the Global Porphyria Advocacy Coalition (GPAC) is Kika Shabot, who is also the Director of the Porphyria Society of Mexico (Sociedad Mexicana Para Porfiria). With the help of Dr. Ronny Kershenovich and some patient advocates, Kika accomplished the impossible in Mexico.

Armed with nothing but sheer determination to help patients receive proper care, they launched a grassroots awareness campaign. With no financial assistance, they spread their message on social media, radio and television. They spoke with two senators, who are now key figures in planning a presentation on rare diseases to the Mexican Congress. Thanks to their hard work, Porfiria is being recognized as a rare disease in Mexico, and Normosang (Panhematin) was reestablished as the obligatory medicine for acute porphyrias.

Congratulations, Kika and Dr. Kershenovich, for your dedication to making treatment accessible to patients. Also Congratulations, Kika, for your new role on GPAC!

Porphyria Society of Mexico's Kika Shabot is the new Vice President of the Global Porphyria Advocacy Coalition.

Italy's New Advocacy Org

DR. FRANCESCA GRANATA

Francesca Granata, PhD, is leading a new porphyria patient organization in Italy. VIVI Porfiria (at **viviporfiria.it**) brings new resources to Italians. "The name 'Vivi Porfiria' represents the essence of our voluntary association, born from the commitment of six patients," Dr. Granata says. Their goal is to create a strong, supportive network between patients, doctors, and family members, with information on specialists and treatment centers, and organizing events and meetings. They also help with diagnosis, access to treatments and education. Stay tuned for Dr. Granata's interview on the *Rarely Discussed* Pamlona edition!

Here's to these Global Leaders

SYLVIE LE MOAL

Sylvie Le Moal of the Association Française des Malades Atteints de Porphyries, is creating an international camp for EPP kids. The APF is collaborating with her on this incredible project. Contact Sylvie at **Association.porphyries@gmail.com** or visit **porphyries-patients.org.**

EILEEN HUDSON

Director of the Chile Porphyria Association, Eileen Hudson, presented to the National Assembly of Chile on behalf of the country's porphyria patients. Eileen's created social media groups for patients in Chile and collaborated with the APF to bring educational events to the community.

Brazil's Powerful Video

ASSOCIAÇÃO
BRASILEIRA
DE PORFIRIA
ABRAPO

The Associação Brasileira de Porfiria (ABRAPO) at **porfiria.org.br** has created one of the most powerful videos we've ever seen! Under Ieda Bussman's guidance, the ABRAPO is becoming one of the strongest patient advocacy groups in the world. You don't need to speak the language to understand this video's impact. You don't need to speak the language to understand this video's impact. View it at **campanha-porfiria.org.**

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.

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Why donate to the APF?

We've put patients first for over 40 years. APF staff work on nights and weekends to help people worldwide find answers to their painful symptoms.

Your donation helps us provide doctor packets filled with pertinent information for newly diagnosed, add to a growing database of 6,000 treating physicians worldwide, support a physician education program, maintain a database of safe and unsafe drugs and more!

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

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

THANK YOU!

UPDATED CONTACT INFORMATION?

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

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3475 Valley Road NW Atlanta, GA 30305

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