AMERICAN PORPHYRIA FOUNDATION

WINTER 2024 **NEWSLETTER**

IN THIS ISSUE

HAPPY HOLIDAYS, MERRY CHRISTMAS,
AND BEST WISHES FOR THE NEW YEAR!

May your lives be blessed with happiness
and good health now and throughout the New Year.
It's our pleasure to serve you.

FROM APF STAFF:
Nicole Castellano, Imani Thomas, Paxton Cumming, Amy Burke,
Nadene Lundmark, Yvette Strange, Carol Hughes, and Andrew McManamon

AND THE APF BOARD OF TRUSTEES:
Paul Sticker, Warren Hudson, Ron Polly, Diana Ijames, James Young, and Desiree Lyon

APF Podcasts from the ICPP

The International Congress of Porphyrins and Porphyrias (ICPP) was held recently in Pamplona, Spain, and the **APF's Rarely Discussed podcast** was there. Executive Director Nicole Castellano and Podcast Producer Andrew McManamon took advantage of world renowned experts, disease advocates, health providers, and researchers in the porphyria community by inviting them on the show.

Listen on your favorite podcast app. New episodes air every two weeks. If you like what you hear, please leave a review! To be a guest on *Rarely Discussed*, contact the APF at **general@porphyriafoundation.org.**

Listen to APF podcast specials recorded at the recent ICPP in Spain.

ICPP—an Enormous Success!

Starting with a Patient Day in Spanish and English, followed by the Scientific Agenda, the Congress was an enormous success. In Pamplona, Spain, attendees were surrounded by beauty, Spanish history and a welcoming atmosphere. ICPP was hosted by the ICPP President, Dr. Antonio Fontenelles, Spanish Porphyria Society President, Fide Miron, and IPNET President, Dr.Sverre Sandberg. The agenda included sessions such as "Heme in Pathophysiology Current Therapies in Acute Porphyria," "New Insight and Potential Treatment in EPP," "Emerging Therapies in EPP," and "Rational Approaches to Diagnosis and Care in the Acute and Cutaneous Porphyrias."

Laura Shares EPP Experience and Poem During ICPP

Laura is an inspiring 17-year-old from Germany, who shared her erythropoietic protoporphyria (EPP) journey via video for the ICPP's Patient Day. Diagnosed at age three, Laura said her life was defined by pain and suffering, as well as the isolation that is often an outcome of living with EPP.

Laura's had the good fortune of being on the SCENESSE® trials that are ongoing for youth in Europe. Her story moved attendees to tears, both from her life-changing experience on the medication, and for all the children still living in the dark.

Laura shared an intimate poem about the challenges of living with EPP. Then, she called on the community to advocate for treatment and research to help people with EPP.

There are things each of us can do, as a community, to spread the word about diagnosing and treating EPP. You can start by watching and sharing **Laura's presentation online.** She will be an upcoming guest on APF podcast *Rarely Discussed*—stay tuned!

Laura lives with EPP in Germany and shared her experience at the ICPP.

Watch Laura tell her EPP story.

Desiree Lyon Receives Top Awards at the ICPP

APF retired Director Desiree Lyon has achieved more FIRSTS in global porphyria history! Desiree received two prestigious awards at the ICPP congress. She received the Lifetime Achievement Award and was inducted as an Honorary Member of the International Porphyria Network (IPNET) for 44 years of devotion to patients and research, including co-founding the APF, establishing the Protect the Future program to train new experts and educate physicians and pioneering the Orphan Drug Act of 1983 gaining Panhematin as the first Orphan Drug. In her career, she helped establish thirteen international advocacy organizations and provided education to patients in over 120 countries. Fide Mirón, President of the Spanish Porphyria Association and co-host of the ICPP presented the Lifetime Achievement Award to Desiree, with thanks from the Global Porphyria Advocacy Coalition (GPAC) and the many patients and physicians attending the ceremony.

President Dr. Sverre Sandberg and Board member Michael Badminton awarded Desiree as an Honorary Member of IPNET. She is the first non-medical honoree of this award. Desiree was among the distinguished company of fellow recipients Dr. Peter Meisner of South Africa and Dr. Rafael Salamanca of Spain.

2024 Patient Day in Italy

On Saturday, November 23rd, the first Italian Patients' Day was held at the Palazzo della Regione Lombardia in Milan. The newly formed Vivi Porfiria group gathered physicians, patients, and caregivers for a day of sharing.

ASSOCIAZIONE VIVIPORFIRIA

The meeting was a major collaboration by the Vivi Porfiria team:

- Dr Francesca Granata, Presidente
- Valentina Rampin, Vice Presidente
- Giulia Schiavina, Segretario
- Cecilia Croce, Tesoriere
- Giulia Gilli, Consigliere
- Danilo Giorgio Romano, Consigliere

Learn more about Vivi Porphyria at **www.viviporfiria.it.**

Porphyria Scientific Findings

The porphyria community is fortunate for the many researchers and scientists worldwide who study this group of diseases. Even if you receive care at a research hospital, it's helpful to keep up on scientific breakthroughs and the latest trends.

The following nine articles were recently published and can be accessed online using Google Scholar. Share with your doctors and caregivers!

1. **Kluijver, Louisa G., et al. "The effects of cholecalciferol and afamelanotide on vitamin D levels in erythropoietic protoporphyria: a multicentre cohort study." British Journal of Dermatology 191.3 (2024): 357-364.**
2. **Jericó, Daniel, et al. "Exploring current and emerging therapies for porphyrias." Liver International (2024).**
3. **Pischik, Elena, et al. "Long-term complications in acute porphyria." Liver International (2024).**
4. **Balogun, Oluwashanu, and Kari Nejak-Bowen. "Understanding Hepatic Porphyrias: Symptoms, Treatments, and Unmet Needs." Seminars in Liver Disease. Thieme Medical Publishers, 2024.**
5. **Badminton, Michael N., et al. "From chemistry to genomics: A concise history of the porphyrias." Liver International (2024).**
6. **Chaiyabutr, Chayada, et al. "Porphyria cutanea tarda in Scotland: underlying associations and treatment approaches." International Journal of Dermatology (2024).**
7. **Riera-Mestre, Antoni, et al. "PICO questions and DELPHI methodology for improving the management of patients with acute hepatic porphyria." Revista Clínica Española (English Edition) 224.5 (2024): 272-280.**
8. **Pischik, Elena, et al. "Long-term complications in acute porphyria." Liver International (2024).**
9. **Polańska, Adriana, et al. "Afamelanotide in protoporphyria and other skin diseases: a review." Advances in Dermatology and Allergology/Postępy Dermatologii i Alergologii 41.1 (2024).**

APF at the Liver Meeting

In November, the American Association for the Study of Liver Diseases hosted its annual conference, The Liver Meeting, in San Diego. APF staff and volunteers represented needs of the patient community among medical experts in hepatology and liver care.

From left: Dr. Bruce Wang, APF Director Nicole Castellano and Recordati representative Daniel Cemulini at the recent Liver Meeting in San Diego.

New Guidelines for Givlaari

A new scientific article is very helpful for those people who are on Givlaari but are experiencing intermittent break through attacks. Because Givlaari lowers PBG, some doctors do not want to treat the attack, despite the patient's insistence that they are indeed suffering an acute attack. Porphyria experts now advise patients on Givlaari receive urgent care for symptoms of acute porphyria attacks. Hemin 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** and provide your doctor with the following article.

If you're on Givlaari and experiencing breakthrough flares, share new care recommendations with your provider.

Moghe, Akshata, et al. "Acute hepatic porphyrias: Recommendations for diagnosis and management with real-world examples." *Molecular Genetics and Metabolism* 140.3 (2023): 107670.

CUTANEOUS PORPHYRIA

What is Pseudoporphyria?

There's a disease that looks like porphyria cutanea tarda (PCT), and acts like PCT, but is *not* PCT.

When Gregory Prosser visited his doctor in 2009, he needed blood pressure medication that wouldn't give him side effects. The doctor prescribed hydrochlorothiazide, but when Greg took it, he started itching badly. Then, blisters appeared. Alarmingly, the outer layer of his skin began to pull away. The itching got worse, and his blisters increased in size, but his physician couldn't determine the cause.

In 2015, his new dermatologist identified the problem as "pseudoporphyria" and gave him cortisone, which helped considerably. At first Gregory thought pseudoporphyria was a type of porphyria like PCT, but quickly learned otherwise.

Whereas porphyrias are metabolic disorders of heme synthesis. Pseudoporphyria describes a rare photosensitivie condition that clinically and histologically mimics PCT without demonstrating porphyrin abnormalities. Pseudoporphyria has been reported in patients with chronic renal failure treated with and without hemodialysis and in people with excessive exposure to ultraviolet A (UV-A) by tanning beds. The cause of pseudoporphyria is due to drugs that interact with sunlight and cause a phototoxic reaction.

If a serum/plasma porphyrin assay is negative, the patient does not have a true porphyria. If the serum/plasma porphyrin assay is unavailable, erythrocytes, urine, and stool may be evaluated for abnormal porphyrin levels. However, such evaluations can represent a diagnostic challenge to complete for patients who may be anuric in the setting of end stage renal disease. At times, people contact the APF thinking their pseudoporphyria is an acquired type of porphyria. Hopefully, this article clarifies that question and this condition.

ACUTE PORPHYRIA

The Difference Between AHP Treatments

Givlaari and Panhematin are two medications for the acute hepatic porphyrias (AHP), including acute intermittent porphyria (AIP), variegate porphyria (VP), and hereditary coproporphyria (HCP). They are both approved to treat acute attacks and symptoms.

Panhematin (hemin for injection) is a approved to treat AHP attacks, consult a specialist about prophylactic use.

PANHEMATIN (HEMIN FOR INJECTION)

Here's how it works: Panhematin provides a form of heme that regulates the imbalance in porphyrin production, reducing porphyrin levels and alleviating symptoms.

Administration: A heme derivative, the medication is administered intravenously. Precise dosing is important.

Effectiveness: Panhematin has been used for over 30 years. It was the first medication approved by the 1983 Orphan Drug Act.

Side Effects: Injection site reactions, nausea, headache, fever, and vein inflammation are side effects. Physicians must monitor iron/ferritin levels. Annual liver imaging is recommended.

VS

GIVLAARI® (givosiran) injection for subcutaneous use 189 mg/mL

Givlaari (givosiran) is a subcutaneous injection approved to prevent AHP attacks and symptoms.

Here's how it works: Givlaari is an RNA interference (RNAi) therapeutic that reduces hepatic aminolevulinic acid synthase 1 (ALAS1) levels to decrease the production of toxic porphyrin precursors.

Administration: An initial subcutaneous injection followed by maintenance doses at specified intervals.

Effectiveness: Clinical studies have shown Givlaari can reduce the frequency of AHP attacks.

Side Effects: Injection site reactions, nausea, and headaches are side effects. APF Members have reported hair loss and extreme weakness/fatigue. Homocysteine levels, as well as liver and kidney health should be monitored.

A Holiday Survival Guide

I can't scroll through Instagram or turn on the TV this time of year without coming across "holiday hacks," tips and tricks to make this season a little less stressful. None mention the added challenge of managing a chronic illness.

The first few years I lived with AIP, I didn't prepare well for the holiday season. I told concerned loved ones I'd get better soon, because that was emotionally easier than trying to explain an incurable disease I hadn't begun to accept. Additionally, I needed to better evaluate how I spent my time and conserved my energy, but I wasn't ready to admit that I couldn't do it all.

I've never heard of a Hallmark movie about someone like me. It's not that there's no societal expectation for those navigating the holidays with a rare and invisible illness; it's that the notion of doing anything other than partaking in the festive grind machine is shunned. There's already extra pressure around the holidays, and chronic disease only complicates things further.

As I hung greenery over my front door and strung lights on the tree, I got to thinking about how I try my best every year to participate in festivities in ways that don't compromise my health and well-being. Here's my holiday survival guide for people with chronic illness.

1. PACE YOURSELF.
My body doesn't do well if I stack activities without rest in between. But we don't have to say no and miss events entirely. Try going for a shorter amount of time.

2. SET EXPECTATIONS.
When I make plans with others, there's only a 60% chance that I'll feel well enough to hang out on that day. My friends know that, but the family members I see once a year don't. As hard as it can be to acknowledge our limitations, it's crucial to clearly communicate our realities, especially during the holidays.

3. SPOT CLEAN.
Porphyria doesn't have to stop you from throwing parties. Instead, create some boundaries for yourself. Focus on clearing clutter, but don't worry if there's a little dust in the corners.

If you're hosting this year, save your energy for friends at the event; don't spend it all on the cleaning beforehand.

4. EAT CONVENIENTLY.
Big, festive meals are part of what makes the holidays special, but working in the kitchen can zap our precious energy. There's no shame in buying pre-made food; 'tis the season for convenience!

Prep what you can in advance or get catering. Most importantly, ask for help and be open to receiving it.

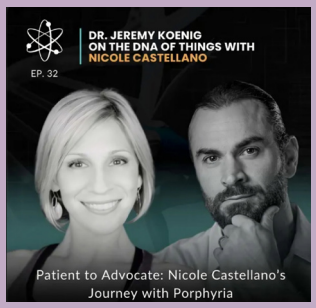
5. PUT YOUR HEALTH FIRST.
Culturally, we're taught to strive for a Folgers-commercial, Christmas-morning feeling. Anything less and the stress sets in. Unlike some of our loved ones, we can't cope with a glass of wine. We also can't afford to lose a night of sleep or miss a meal. This tactic is the most important on the holiday survival guide.

6. NOURISH YOURSELF.
Just by living with porphyria, we're resilient. Not only do we survive attack after attack, but we continue to thrive and grow. We all have ways to find calm amid the chaos of our symptoms. Whether it's reading a book by the fire, watching a holiday movie with your fur baby, or calling your best friend to laugh, make a list of the ways you care for yourself and lean on it. Regardless how you observe the season, find compassion for your body first.

ABOUT THE AUTHOR
Claire Richmond
Writer, Lives with acute porphyria

A Porphyria Podcast Episode

Check out APF Director Nicole Castellano on **The DNA of Things podcast** with Dr. Jeremy Koenig. Nicole shares intimate details of living with AIP. The episode also highlights the importance of accessible testing, and evolving roles of technology and AI in transforming rare disease care. Find this and all *The DNA of Things* episodes on your favorite podcast app.



APF Director Nicole talks porphyria on a recent The DNA of Things podcast.

DNA Testing

The APF will continue connecting the community with free genetic testing through Prevention Genetics and the Alnylam Act Program. DNA testing is critical to the diagnosis of porphyria, but the expense of genetic testing is a barrier to patients and family members obtaining important medical information. **Contact the APF** to receive a free DNA test form.

Dr. Michel DeBakey and1,000 Flaming Swords

One of my descriptions for symptoms of acute porphyria is the pain of "a thousand flaming swords." Interestingly, this short phrase has found its way into graphics about acute porphyria, and patient stories around the world. Unfortunately, similar descriptions are believed by some physicians to be extreme exaggerations. Patients using such descriptors are often viewed as being overly dramatic, and ignored. Although my thousand burning, searing swords description was closer to the truth than others, I too was ignored, until a chance visit from an old friend. Here is how my life changed.

I have AIP. Like many individuals who suffer from rare diseases, I endured a decade of dreadful life-threatening attacks before properly diagnosed. My circuitous route to diagnosis was peppered with many emotionally devastating accusations by medical professionals. I was called a "drug seeker," a "hypochondriac," and other embarrassing names that found their way onto my medical record.

After years of being cast in such a demeaning light, I was finally diagnosed. My attacks continued to rage until I received Panhematin, which that helped quell the horrendous pain.

World-renowned heart surgeon, Dr. Michel DeBakey, helped physicians understand the severity of porphyria pain, which validated generations of patients once seen as being "dramatic."

During one of my particularly painful AIP sieges, I was visited in the hospital by a friend, the famous heart surgeon Dr. Michel DeBakey. He'd seen me in numerous difficult attacks, and this time he was accompanied by a group of young doctors making rounds. Upon entering my room, Dr. DeBakey asked if I would help his residents learn about porphyria. Despite being in writhing pain, I quickly agreed to answer any questions.

Dr DeBakey began by explaining the acute porphyrias and included why it was such a painful disease. I was surprised when he asked me to share my personal experience with porphyria pain. After using a few general terms like, "agonizing, unspeakable and unbearable," I ended my short presentation by likening the pain to that caused by "a thousand flaming swords." I could tell by the expressions on the assembled doctors' faces that my description did not land well.

To my surprise, Dr. DeBakey came to my rescue, stating that most acute porphyria patients are not believed because their complaints appear over-exaggerated, and that my flaming swords description was a prime example. He said, "In an attack, some patients have pain so severe that is feels like having surgery—without anesthesia!" I was initially shocked by his seemingly outlandish example, but for the first time I felt validated.

After Dr. DeBakey's stern admonishment to the assembled medical professionals, a few of them apologized to me and asked to know more. I also learned a great deal, especially that it takes the best educational tools and resources to advance our message about porphyria and the pain. We cannot always have a famous doctor at hand to plead our case, but we can make sure that what we provide to our medical team is top-notch material. That's why we include a pain letter from a porphyria expert in the APF's free doctor packet.

Do you have a story like Desiree's experience with Dr. DeBakey, or an analogy you use to describe your porphyria pain to others? We want to feature them! Email **general@porphyriafoundation.org.**

REQUEST A FREE DOCTOR PACKET!

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

ABOUT THE AUTHOR

Desiree Lyon
APF Founder and Retired Director

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

Connecting the Porphyria Dots

Many aspects of daily living were oddly taxing until well into my fifties, when I learned of my acute porphyria diagnosis. Now I can look back on my life and identify significant attacks.

Symptoms started my senior year of high school, when I missed a full quarter's worth of classes due to unidentified medical issues. I kept my grades up because I had so much time to read. Then I started nursing school, but found I simply did not have the energy for the tasks. I changed to a liberal arts degree, met my husband, and was married at age 19. I got pregnant at 21, and had a baby boy after some difficulty. After, I had a hideous episode of pain and general illness. As I recovered, I became pregnant with our daughter. It was a difficult pregnancy, and she was delivered 6 to 8 weeks early. I was advised by a team of hematologist and rheumatologist that I should not become pregnant again. However, my gynecologist was supportive and I soon became pregnant with our second daughter. She was my first normal pregnancy, but a few hours after delivery I began to have severe medical issues.

My final major episode came about age 50 with menopause. I was quarantined in the hospital with sores and pain in my gut and legs so horrible that I was on a morphine drip for 2 weeks. Tests were done, and "porphyria" was mentioned. I was sent to a teaching hospital to see a doctor who had treated one AIP patient. He doubted the diagnosis because he'd heard of it mostly affecting alcoholics. However, his resident persisted in getting the test ordered and thankfully, I got an answer. I continued to be hospitalized with more minor episodes during menopause.

I retired early at age 60. I read and enjoy riding horses in my spare time. In search for a physician referral for my persistent pain, I contacted the APF. I was put in touch with a doctor who just happened to be a distant cousin I'd met at a family reunion when we were both children! He told me we all carried genetic markers that made porphyria very likely. I'm grateful to the APF's resources and contribution to advancements in porphyria treatment.

Read my story in its entirety in the patient stories section. You can find me as "Marsha Prock" on Facebook, I'd love to hear from anyone who can relate to my story.

ABOUT THE AUTHOR

Marsha Prock
Lives with
acute porphyria

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

IN MEMORY

David Mollura for
Rev. Darlene M. (Fletcher) Bishop

Elly Hensman for
Rev. Darlene M. Bishop

Dianne Fletcher for
Rev. Darlene M. Bishop

Janet Jordan for
Rev. Darlene M. Bishop

Cindy & Michael Donahue for
Rev. Darlene M. Bishop

Montgomery School for
Rev. Darlene M. Bishop

Erin Paradis-Alshefski for
Rev. Darlene M. Bishop

Carolina Tamayo for
Maria Libia Amaya Alvarez

Pamela Marin for **Eric Lifschitz**

Desiree Lyon and Nicole Castellano for **Eric Lifschitz**

Desiree Lyon, Pax Cumming, Imani Thomas, Amy Burke, Nadene Lundmark, Carol Huges, and Yvette Strange for **Mila Castellano**

IN HONOR

Michael Nichols for **Katayuki**

Audra Dehan for **Barbara Haskell**

Charles Davis for **Ginger Davis**

Geneva Wright for
Rev. Darlene M. Bishop

Nhi Chau for **Rev. Darlene M. Bishop**

Bonnie Niglio for **Nicole Castellano**

Sharon Koch for **Jagger and Jake**

The APF community remembers Mila, the viszla.

Honoring Mila's Memory

Last month, APF Director Nicole Castellano tragically lost her beloved canine companion Mila "La Dolce Vizsla." The animal lovers in our community understand how much emotional support and healing a pet provides, particularly when we feel alone in the pain of our porphyria. Mila was a special member of the APF family, and the inspiration for the pet calendar. Our deepest sympathies to Nicole as we honor Mila's memory.

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.

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.

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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