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Recapping Porphyria Awareness Week 2023



was "Spotlight Porphyria."

Dr. Roy Smith, hematologist at the University of Pittsburgh and



Dr. Roy E. Smith kicked off PAW with a Zoom presentation on April 15, 2023.



CLICK HERE
to watch
Dr. Smith's
presentation

Porphyria Awareness Week (PAW) 2023 took place April 15-22. The theme

renowned porphyria expert hosted an educational Zoom to kick off the week. Mayra Martinez, Megan Parrish and Sharon Dill shared experiences living with acute porphyria. Over 200 patients and physicians were also involved. Thanks to all!

In honor of PAW, Recordati Rare Disease launched a click campaign that donated \$5 to the APF for every click. Members raised \$5,000! Thanks to Recordati for supporting our education programs.

Thanks also to Mitsubishi Tanabe USA and Clinuvel for assistance with PAW



Member George Hodder rocks his purple in an APF cycling shirt. "Porphyrus" is Greek for purple, and it's the color for porphyria.

activities. Finally, thanks to all of YOU members for your unwavering support. You are the APF.

Here's the week's activities:

Satisfy the Soul Sunday: We encouraged our community to reflect on their blessings.

Motivational Monday: We asked for tips to cope. We shared the [Two of Me](#) documentary, [Lifetime Network's The Balancing Act](#) and Nicole Castellano's [Suncoast News Network interview](#).

Talk Tuesday: The community showcased their personal journeys during the *Let's Talk Porphyria* Zoom hosted by Nicole Castellano.

Wear A Hat Wednesday: We wore hats for kids with EPP (and all photosensitive porphyrias)!

Tell Your Story Thursday: We asked the community to share stories about life with porphyria.

Find a Friend Friday: We featured the Porphyria Partners Program (PPP), designed to counteract the loneliness felt in our community. (More on page 2.)

Show your Color Purple Saturday: We encouraged our community to wear purple, and social feeds were full of porphyria-themed selfies!

THE APF IS PATIENTS-FIRST AND 17,500 STRONG

We are growing faster than ever in our 40-year history! Our patients-first policy draws patients and doctors to the APF for help from across the country and around the world.

Global Porphyria Day

This year's Global Porphyria Day was April 19, and groups around the world observed the date. The Brazil Porphyria Foundation hosted a series of activities and educational projects. The Canadian Porphyria Association lit the capitol with purple, and Alnylam Pharmaceuticals threw a party.



Alnylam Pharmaceuticals celebrated Global Porphyria Day.

2023 APF Presidential Award

Nicolas Frias (pictured, right) was awarded the 2023 APF Presidential Award for developing the mobile app porphyriadrugs.com. He developed the site years ago, and continues to update and improve it.

Anyone using the old version can reinstall by visiting porphyriadrugs.com on their mobile device or porphyriadrugs.com/mobile.



Nicolas developed the site gratis because acute porphyria is in his family, and he knows the importance of safe/unsafe drugs.

Porphyria Partners Program



The APF initiated Porphyria Partners Program (PPP) during the Covid pandemic to counteract isolation

and loneliness. PPP worked well, so we're expanding, creating an ongoing program.

Debra Knapp, who has HCP, will lead the PPP. Debra was an attorney who thoroughly enjoyed her full workload and active life until she began to experience horrific acute attacks. Like many patients, the cause was a mystery. Her disease became so debilitating she could no longer

work. She was eventually diagnosed, but remains intermittently ill.

If you need a "porphyria partner" or want to help others, [email general@porphyriafoundation.org](mailto:general@porphyriafoundation.org) or find Debra Knapp on Facebook.



Debra Knapp devotes some of her free time to porphyria advocacy through the PPP and serving on the APF's Member Advisory Board.

Spring EPP Patient Zoom

Recently, the APF and Canadian Porphyria Association hosted a Zoom for erythropoietic protoporphyria (EPP). Dr. Karl Saardi presented and answered questions. Loren and Craig De Mase told their story as couple (patient and caregiver) managing EPP, and

Temperance Stala shared as a child with the disease. Michelle Capon, Canadian association vice president, explained what life is like without treatment.

Dr. Karl Saardi is a dermatologist at George Washington University Hospital and an expert in EPP.



Pharma Friends: Nancy Bayerlein

Nancy Bayerlein is the Senior Director of Patient Engagement and Support for Mitsubishi Tanabe Pharma America. She's perfectly suited for her position, as she has a huge heart for patients. Nancy worked in patient advocacy for 20 years before landing at MTPA. Their company-wide ideology is the same as the APF—patients first! Her role is especially gratifying as she

helps define and fulfill unmet needs of patients. It allows her to meet and work with advocacy groups like the APF, as well as other non-profit organizations that have a focus on healthcare. The favorite part of her job is the people, who are interesting, bright and creative, and together they create new programs to help patients and care partners.

When Nancy is not working, she loves to travel with her family and is planning to celebrate her daughter's university graduation with an upcoming trip to Italy and Greece. Nancy also devotes herself to animals of all kinds. She particularly enjoys riding and driving Clydesdale horses. Nancy lives on top of a small mountain in New Jersey and loves the outdoors, where she is surrounded by the beauty of the landscape and what she describes as, "lots of critters," bear, deer, and even bobcats.

Nancy's equine therapy volunteer work for developmentally disabled children, combines her love of people and horses. Equine therapy helps children enhance their muscle control, self-confidence and a sense of accomplishment.

Editor Note: We look forward to the Mitsubishi trial results.



Horses are a passion for MTPA Director of Patient Advocacy Nancy Bayerlein. Here's a photo of her with beloved horse, Luke.

New T-Shirt Design is on Fire

Patrick Petersen of Oklahoma City, OK is excited about his \$100 prize for his "por-FIRE-ia" design.

Patrick is a self-taught artist with a large online following. Check out all of his animated videos on his [YouTube channel @NighttimeOfficial806](#).



My name is Pat Petersen, and I am 14 years old. I enjoy drawing, making animations, creating new characters, and writing stories. My mom has AIP, and I often help her out and pray for her during attacks. So many times, I've heard her say that her spine was "on fire," or that it felt like there were volcanic rocks grinding against each other in her abdomen. This inspired me to make the "por-fire-ia" dragon for the t shirt contest. I am glad it was chosen, and it helps more people learn about porphyria.



PAT PETERSON,
artist and son of an
AIP Super Mom



CLICK HERE
for this and
other stylin'
APF apparel at
the online store!

An Inspiring Acute Porphyria Video

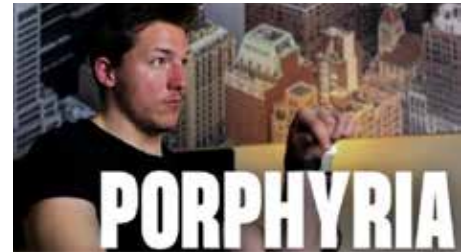
During a family vacation in 2018, Andrew was enjoying some of his favorite activities: fishing, sports and off-road biking. The fun stopped when he woke up with severe pain he likened to a knife being drawn through his abdomen. His every effort to quell the pain was futile. Finally, he went to a small town hospital nearby where he was tested for the typical causes of abdominal pain. When nothing showed up, he was told the attack was in his head. A few days later, he repeated the scenario, but to no avail. Once home from vacation, he

visited his primary care provider, who hospitalized Andrew and promptly tested him for acute intermittent porphyria (AIP). Andrew didn't have years of a diagnostic nightmare, thanks to a doctor who knew to think of porphyria.

During PAW, Andrew posted about his experience with AIP on YouTube, and **the video** was shared widely among the porphyria community. Recently, Andrew agreed to work with the APF on more videos. Stay tuned to the APF social channels for more videos.



[CLICK HERE](#) to watch Andrew's video on the APF website!



After diagnosis, Andrew reached out to the APF and began advocating for porphyria on his YouTube channel.

CUTANEOUS PORPHYRIA

Living Passionately Despite CEP Challenges

Fide Mirón is president of the Spanish Association of Porphyria, and vice president of the Spanish Federation of Rare Diseases. She lives with congenital erythropoietic porphyria (CEP) and recently shared how illness complicated her childhood, and how it impacts her today.

I graduated with a degree in social work from the University of Alicante, which excites me because I can also use my life experience to be a social transformer. I am passionate about giving motivational conferences that are aimed at students, health professionals, organizations, and work teams whose main theme is motivation and, above all, people with the desire to grow. Due to my extensive experience, I like to

contribute to seeing the world with a new perspective and self-improvement. Joy and the desire to live are the weapons I use to focus on what is most important—living passionately.

Having porphyria has made me feel very alone in my life health-wise. I am grateful now for the great opportunity to accompany other patients on their journeys, for being involved in the first bone marrow transplant case in Spain, and for seeing its fantastic results for porphyria and other diseases. Another honor has been meeting the researcher Oscar Millet and his entire team, seeing how the research progresses, and with it, the hopes of changing the future for new patients.



Fide Mirón is a global leader in porphyria and lives with CEP. She recently shared her story with the APF.



[CLICK HERE](#) to read Fide Mirón's story on the APF website!

Blood Drive for CEP Raises Awareness

The Covid pandemic hit hard and impacted the availability of blood. This put some, like people with porphyria, in life-threatening situations.

Amanda and Jose Rivera live in Clovis, CA with their children, Harmoni (age 4) and Mateo (age 1), who each have congenital erythropoietic porphyria (CEP). Harmoni needed blood transfusions weekly, but was forced to wait due to a national blood shortage.

Her parents sprang into action! They launched a blood drive in the San Joaquin Valley with the tagline: Valley People Need Valley Blood! The

Riveras continued holding the blood drive annually. They even have a lifelong code (282C), which anyone in California can use at any time.

Jose learned he was a match to give Harmoni bone marrow. She received a transplant, and it was successful! Now she only needs blood once a month. The Rivera's situation as a whole seem to be improving. Amanda's noticed a reduction in wait time for Mateo, who needs blood transfusions bi-weekly.

For three years, the Rivera's blood drive has been part of PAW! The APF

happily provides materials on CEP to be distributed at the event. Well done and best wishes to the Riveras!



Harmoni (back) and Mateo (front) live with CEP and need blood transfusions. Their parents hosted a blood drive to counteract recent shortages.

Greater than my Circumstances, and my PCT

APF Member Murphy McNutt shares the story of her diagnostic journey, and the APF's role in getting treatment and answers.



MURPHY MCNUTT, who lives with PCT

I consider myself to be a confident, healthy, and strong woman with a whole lot of life to live. Life is beautiful, and I never knew how much I took it for granted until 2016 hit me. I didn't feel like myself at all. Juggling a full-time job and teaching fitness classes, I thought I was doing all of the right things to lead a healthy lifestyle. What could possibly be wrong?

That summer, I misplaced a lab order. Months later at a check up, my doctor said my liver enzymes were very high.

About three weeks later, I discovered a huge, painful blister on the top of my hand. My doctor asked if I'd touched anything that could've potentially infected it, but that wasn't what was going on. Soon blisters popped up all over on the tops of my hands. It was painful and I was mortified at how I looked.

I was overcome with panic, worry and anxiety at my first appointment when, at last, I saw a liver specialist. Despite high porphyrins, the porphyria cutanea tarda (PCT) connection still wasn't made. My life became a revolving door, I was going in and out of a doctor's offices. With each appointment, my hope, confidence, and resilience faded.

I researched my symptoms and eventually discovered the APF, which led me to an expert who finally confirmed PCT. After learning what I needed to be healthy, including phlebotomies and some lifestyle changes, I went into remission. The scars on my hands are a constant reminder that I am greater than my circumstance. I no longer take my health for granted and am a better person because of my PCT.

APF Advocates for People Around the World

The APF serves patients worldwide and has assisted people in over 100 countries. For 25 years, APF Executive Director Desiree Lyon anonymously gave her salary back to the APF so that funding would be available for special patient and physician education programs, including the global program. We are proud of the work we've accomplished worldwide. Most recently, we assisted a young woman in Pakistan, who was in critical condition with AIP, and a young man with CEP. In Egypt, we

helped a young woman with AIP, who had no treatment for her serious attacks. Thanks to help from the APF, she is now greatly improved.

A physician in Costa Rica asked the APF to locate a porphyria expert and information on treatments. A child in Venezuela needed help securing an EPP diagnosis. Nadezda Pejovic wanted to start a porphyria group in Serbia, and the APF is assisting (email general@porphyriafoundation.org to learn more).

The APF maintains a database of 5,500 physicians globally. A patient in Nairobi sought assistance locating a doctor. With friends like Sylvie LaMoal in France and Sue Burrell in the United Kingdom, we provide support to many internationally. Congratulations to the new patient organization Associazione ViviPorfiria! For our Italian friends, please visit www.viviporfiria.it and watch for more news in our Fall newsletter.

APF Director at the Capitol

As an early pioneer of the Orphan Drug Act (ODA) and leader in the movement to secure an acute porphyria treatment called Panhematin as the first orphan drug, APF Executive Director Desiree Lyon was invited to speak at the U.S. Capitol. It's the 40th anniversary of the ODA, and Desiree is one of the influential few who can still remember those changing times. She described life before the ODA, and how the legislation revolutionized healthcare for rare conditions, like porphyria. Patient organizations worked so hard advocating for the ODA, and it was a thrilling achievement to really see it in action.

Forty years ago, there was a scant few drugs to treat rare diseases. Now, there are over one thousand with "orphan drug" status! "Helping

secure the ODA and Panhematin as the first orphan drug is one of the greatest moments of my life," Desiree said.

Paul Stickler, APF board of trustee member, also worked on the first ODA project and served as moderator of the presentation. Thanks to Mike Eging, head of the Rare Action Access Project, for arranging the presentation. Mike also helped establish Panhematin as the first orphan drug.



In April, Desiree Lyon shared how her work resulted in Panhematin becoming the first orphan drug.

Pharma News



Disc Medicine maintains the following EPP research: *Study of Bitopertin to Evaluate the Safety, Tolerability, Efficacy, and PPIX Concentrations in Participants With EPP*. To participate, in the U.S. contact the APF at 866 APF-3635. People in Canada can contact the Canadian Porphyria Association at coordinator@porphyriacanada.com.



Clinuvel expanded its global porphyria programs, including VP trials and treating EPP patients in Canada with Scenesse (Afamelanotide). Scenesse is a prescription implant to increase pain-free light exposure. It's prescribed for adults with a history of phototoxic reactions from EPP. Interested in EPP treatment? Visit www.scenesse.com.

People on Givlaari Still Need Panhematin

By: Claire Richmond

I've had indigestion, period cramps, bloating, constipation, tumors on my ovaries and abdominal surgery. And while painful in their own right, none of those conditions come close to the category of pain elicited by AHP.

Even doctors who see AHP patients regularly don't understand porphyria attack pain. This is incredibly frustrating when I'm in an attack and just want my crisis to be taken seriously. But I can see why this issue

is complicated. Measuring pain on a ten-point scale is impossible, and people in attacks respond differently. It doesn't help that little to no research has been done on porphyria pain.

If there's one thing I've learned about living with AHP, it's that I have to prove myself time and again. Depending on who I see in the emergency room, if I need an urgent infusion of Panhematin (hemin for injection) for a bad attack, it's not enough to have a genetically confirmed diagnosis, or a

demonstrated disease history.

People everywhere with acute porphyria experience these barriers for accessing treatment. But with the advent of Givlaari, a new preventative treatment, yet another obstacle has begun to emerge.



CLICK HERE
to read full story on the APF website!

APF FRIENDS OFTEN GIVE DONATIONS IN MEMORY OR HONOR OF THEIR DEAR LOVED ONES. WE ARE GRATEFUL FOR EACH OF THEM.

In Memory



Kevin, Catherine, and Louie Dolezal in Memory of Susan Young

Jeff Ravitz in Memory of Susan Young

Gary Graff in Memory of Susan Young

Marsha Rivers in memory of Jim Witter

Desiree Lyon in memory of Jim Witter

Julian Dean and Launa Pierce in memory of Jim and Barb Witter

Jess Allen Family in memory of Derek Search

Patrick and Shari Wright in memory of Jim Witter

Roger and Kathy Veal in memory of Jim Witter

Linda and Bob Marino in memory of Theodore Saddic

In Honor

Marcia and Louie Alicea in Honor of Tasha and Kalel Alicea

Lori Hanson in Honor of Ralph Gray

Amy Chapman in honor of her mother, Christie, sister Michelle, and Uncle John who fight AIP daily!

Mike & Rosemary Sieben in honor of Nicole Castellano

The APF staff in honor of all the porphyria patients around the world

Desiree Lyon in honor and thanks for the APF Member Advisory Board



CHEERS

Best wishes to the soon to be Mr. and Mrs. Maggie and David Slusser, who will wed June 3rd in Grand Rapids, MI.

Cheers to Kelsey Heflin and Max Crow on their engagement, they're planning an October wedding.

Congratulations to Elizabeth Brouger on graduating from the University of Georgia.

TEARS

Tears over the passing of James Witter on April 12, 2023 in Tulsa, Oklahoma.

James was an APF member since its inception. His support assisted in physician and patient education.

We most are proud he cared for his daughter, Terri, during her bouts of AIP.

What's New?

Check out www.PorphyrriaFoundation.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.

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Donate to the APF

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

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